S of DNA:

HELPING PATRONS UNRAVEL THE MYSTERY OF GENETIC INFORMATION

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Genetics Overview

Genomic Health Literacy

Genetic Testing

Consumer Health Resources

Ethics & Privacy

All of Us Research Program

NNL…Huh?

NIH
- **National Institutes of Health**
- Nation's research agency

NLM
- **National Library of Medicine**
- World's largest biomedical library

NNLM
- **National Network of Libraries of Medicine**
- Program of the NLM comprised of 8 Regional Libraries (RMLs) and 6 offices

NNLM

U.S. National Library of Medicine
National Network of Libraries of Medicine

U.S. National Library of Medicine
National Network of Libraries of Medicine
Pacific Northwest Region

The mission of NNLM is to advance the progress of medicine and improve the public health by:

- Providing all U.S. health professionals with equal access to biomedical information
- Improving the public's access to information to enable them to make informed decisions about their health

NNLM
https://nnlm.gov/

Genetics in the News

NIH U.S. National Library of Medicine
National Network of Libraries of Medicine

Human Gene Editing Receives Science Panel's Support

Scientists Say They Hope To Create A Human Genome In The Lab

Scientists Use Genetic Engineering To Vanquish Disease-Carrying Insects

Baltimore Ravens to hand out free DNA test kits

Clinical Genetics Has a Big Problem That's Affecting People's Lives

Unreliable research can lead families to make health decisions they might regret.

Mail-Order CRISPR Kits Allow Absolutely Anyone to Hack DNA

Genetic Testing for Athletic Ability

Can genes predict sporting talent?

Opioids: Can a Genetic Test Identify an Addict in the Making?

Genetically Modified Humans? How Genome Editing Works

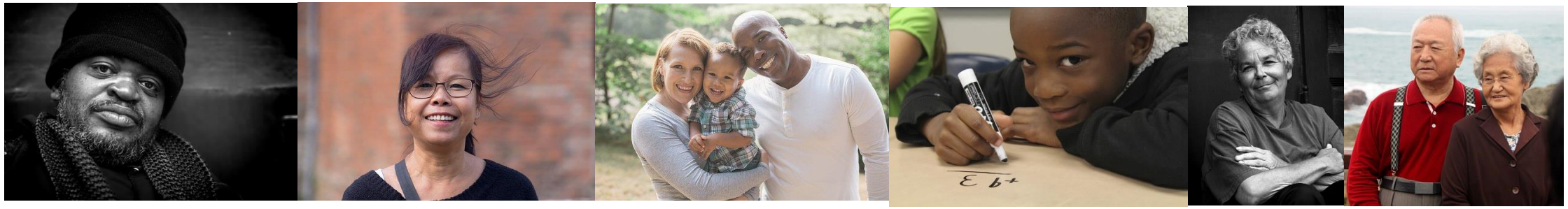
Birth of Baby With Three Parents' DNA Marks Success for Banned Technique

Genomic Health Literacy

Lack biology basics

Lack mathematical concepts

Low health literacy



Definitions

Genomic Health Literacy

- The capacity to obtain, process, understand, and use genomic information for health related decision making.

Genomic Science Literacy

- The knowledge of basic genetics and genomics concepts and processes needed to build conceptual understanding, and the necessary mathematical knowledge to support this comprehension.

Leading causes of death

1. Heart disease: 633,942
2. Cancer: 595,930
3. Chronic lower respiratory diseases: 155,041
4. Accidents (unintentional injuries): 146,571
5. Stroke (cerebrovascular diseases): 140,323
6. Alzheimer's disease: 110,561
7. Diabetes: 79,535
8. Influenza and pneumonia: 57,062
9. Nephritis, nephrotic syndrome, and nephrosis: 49,959
10. Intentional self-harm (suicide): 44,193

CDC FastStats

The Story of You

The Story of You

CATEGORIES OF DISEASES ATTRIBUTED TO GENES

- Chromosomal Diseases
- Monogenic Diseases
- Multifactorial Diseases

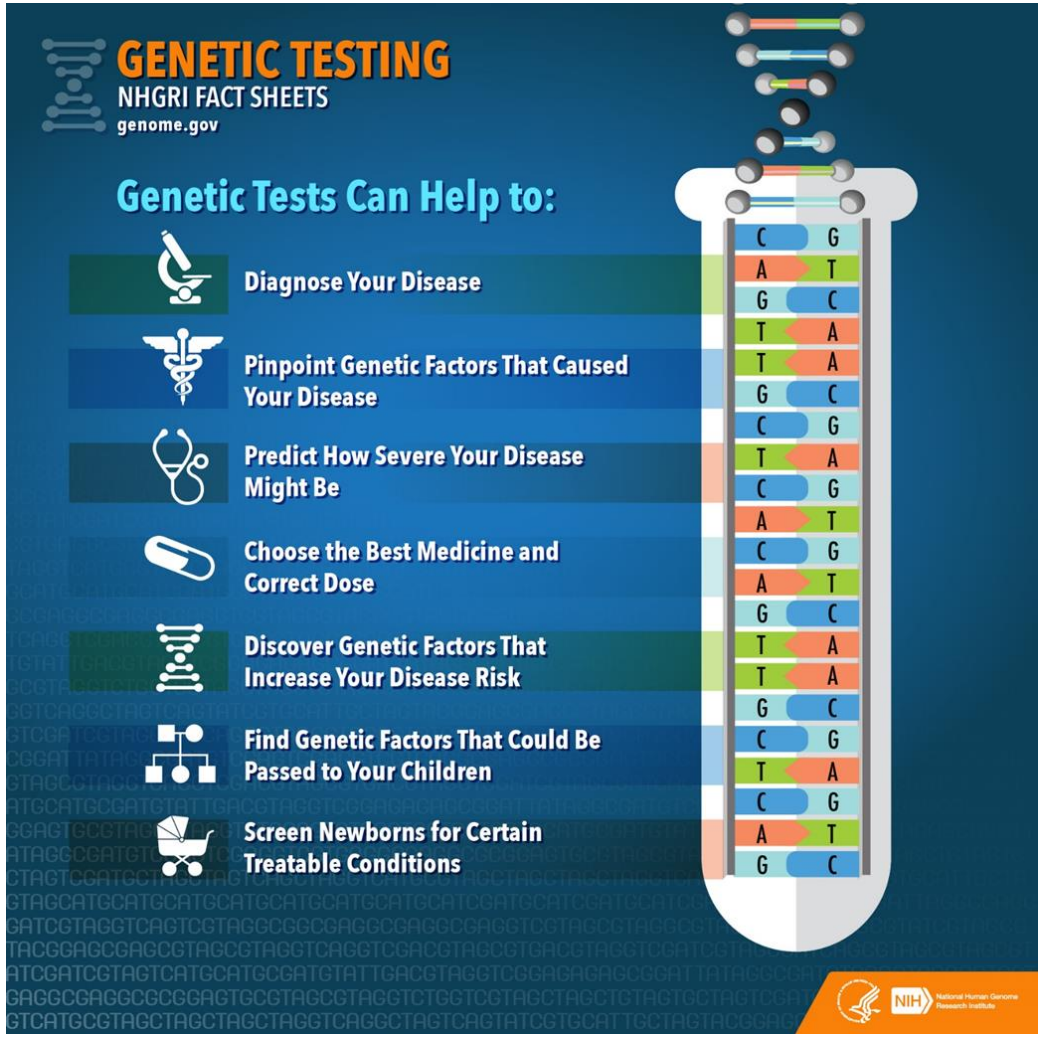
Genetic Testing

INCLUDING DIRECT-TO-CONSUMER

Types of Genetic Tests

- Diagnostic
- Predictive
- Carrier
- Prenatal
- Newborn Screening
- Research
- Pharmacogenetic

Clinical Uses of Genetic Tests



[Genetic Testing]

Jean's Genetic Testing Timeline

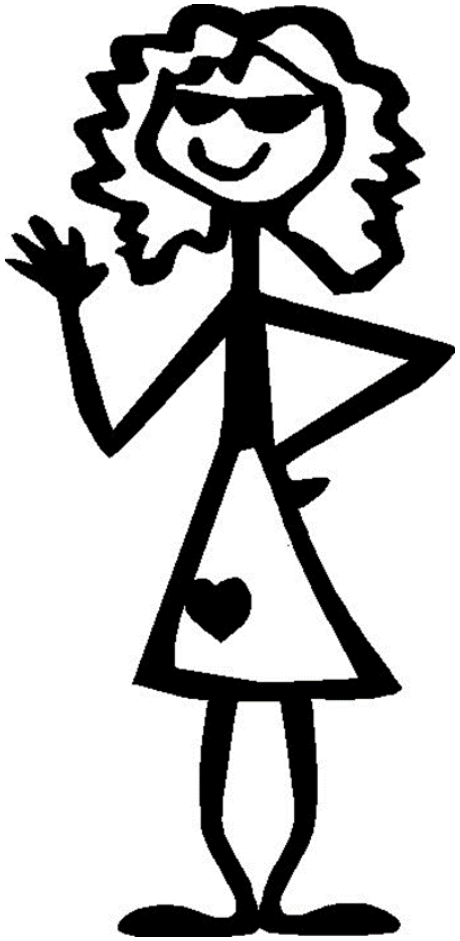
Age 1 day: **newborn** testing for a few serious childhood diseases

Age 30: **carrier** testing (with her partner) before getting pregnant

Age 35: **predictive testing** when sister develops breast cancer at a young age

Age 45 **direct to consumer** genetic testing to investigate ancestry

Age 65 **pharmacogenomics** testing when Plavix wasn't effective



Genetic Testing Results

What genes and what variants did you test for?

- Different tests offered for the same conditions.
- Knowledge always changing.

Might not have enough examples in the database to determine associations between specific variants and specific conditions.

Might not have enough examples of people like you in the database.

Possibility of false positive and false negative results.

BRCA 1 & 2

- Majority of breast and ovarian cancers are not linked to BRCA
- Only 0.2% carry BRCA mutations
- U.S. Preventive Services Task Force recommends that women who have family members with breast, ovarian, tubal, or peritoneal cancer be assessed
- Women who are found to have a family history that may be associated with BRCA1 or BRCA2 mutations should receive genetic counseling and subsequent BRCA testing, if indicated
- Having the mutation does not necessarily mean cancer will develop, but it does increase risk

Lab Tests Online BRCA information
NCI BRCA Fact Sheet

DTC BRCA test

U.S. Department of Health and Human Services

FDA U.S. FOOD & DRUG ADMINISTRATION

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FDA News Release

FDA authorizes, with special controls, direct-to-consumer test that reports three mutations in the BRCA breast cancer genes

Test only reports 3 out of more than 1,000 known BRCA mutations and negative result doesn't rule out increased cancer risk

SHARE TWEET LINKEDIN PIN IT EMAIL PRINT

For Immediate Release March 6, 2018

Release

Español

The U.S. Food and Drug Administration today authorized the Personal Genome Service Genetic Health Risk (GHR) Report for BRCA1/BRCA2 (Selected Variants). It is the first direct-to-consumer (DTC) test to report on three specific BRCA1/BRCA2 breast cancer gene mutations that are most common in people of Ashkenazi (Eastern European) Jewish descent. These three mutations, however, are not the most common BRCA1/BRCA2 mutations in the general population.

Inquiries

Media

Deborah Kotz
301-796-5349

Consumers

888-INFO-FDA

Related Information

- FDA allows marketing of first direct-to-consumer tests that provide genetic risk information
- Commissioner statement on consumer tests that evaluate genetic health risks
- Diagnostic tests

Follow FDA

23andMeBlog

HOME CATEGORIES ALL POSTS

A Major Milestone in Consumer Health Empowerment

March 6, 2018 By AnneW under Health and Traits

We wear pink ribbons. We walk or run for the cause. We have a month for awareness, and yet many women who have a higher risk variant in the BRCA1 or BRCA2 genes remain unaware of their heightened genetic risk for breast and ovarian cancer until a doctor diagnoses them with cancer.[1]

That's a failure.

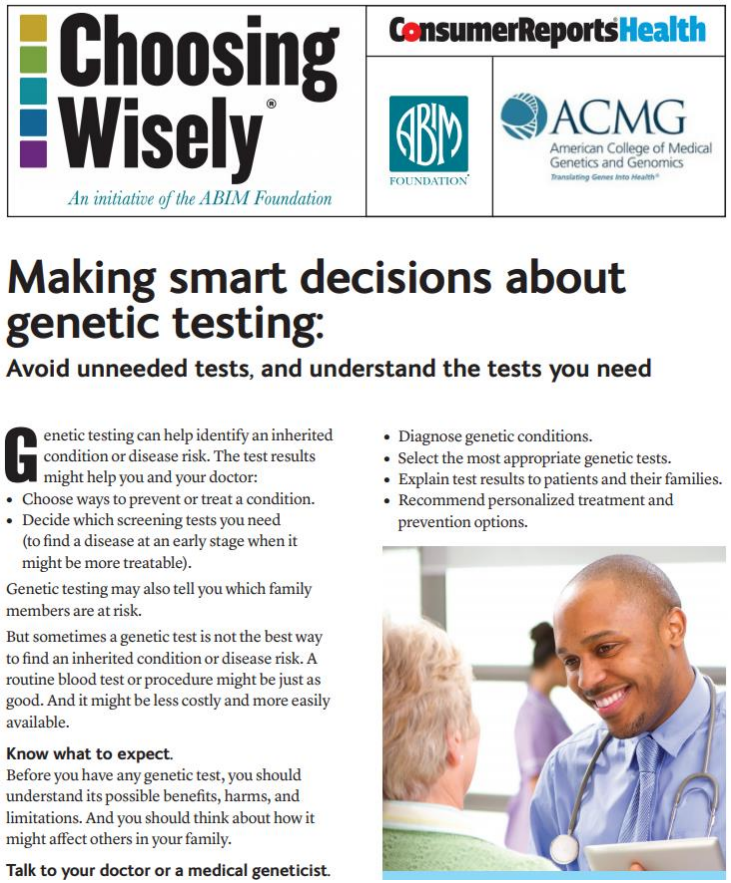
This is not a radical indictment of the American healthcare system; it's just a fact. We are very good at treating illnesses, but not at preventing them.

Under the current system, there are specific guidelines for BRCA screening that limit who has access to BRCA testing. Family history of cancer and Ashkenazi Jewish ancestry are the most common criteria for screening guidelines. However we have seen at 23andMe that many people do not know their family medical history or their ancestry. So,

FDA announcement

23andMe announcement

Genetic Testing- is it necessary?


Choosing Wisely®
An initiative of the ABIM Foundation
ConsumerReportsHealth
ABIM FOUNDATION
ACMG American College of Medical Genetics and Genomics

Making smart decisions about genetic testing:

Avoid unneeded tests, and understand the tests you need

Genetic testing can help identify an inherited condition or disease risk. The test results might help you and your doctor:

- Choose ways to prevent or treat a condition.
- Decide which screening tests you need (to find a disease at an early stage when it might be more treatable).

Genetic testing may also tell you which family members are at risk.

But sometimes a genetic test is not the best way to find an inherited condition or disease risk. A routine blood test or procedure might be just as good. And it might be less costly and more easily available.

Know what to expect.
Before you have any genetic test, you should understand its possible benefits, harms, and limitations. And you should think about how it might affect others in your family.

Talk to your doctor or a medical geneticist.

- Diagnose genetic conditions.
- Select the most appropriate genetic tests.
- Explain test results to patients and their families.
- Recommend personalized treatment and prevention options.

[Choosing Wisely]

Questions to ask:

- Am I in the group at risk and should I get tested?
- If I decide to get tested, what do the results mean?
- What are my treatment options based on results?
- How do I decide on treatment?

Genetic Counselors

- Evaluate family history and medical records
- Assist in making decisions regarding genetic testing
- Identify and interpret risks of inherited disorders, increase the family's understanding of a genetic condition
- Discuss options regarding disease management and the risks and benefits of further testing and other options
- Help the individual and family identify the psychosocial tools required to cope with potential outcomes
- Reduce the family's anxiety

National Society of Genetic Counselors

Direct to Consumer Testing

ancestryDNA™

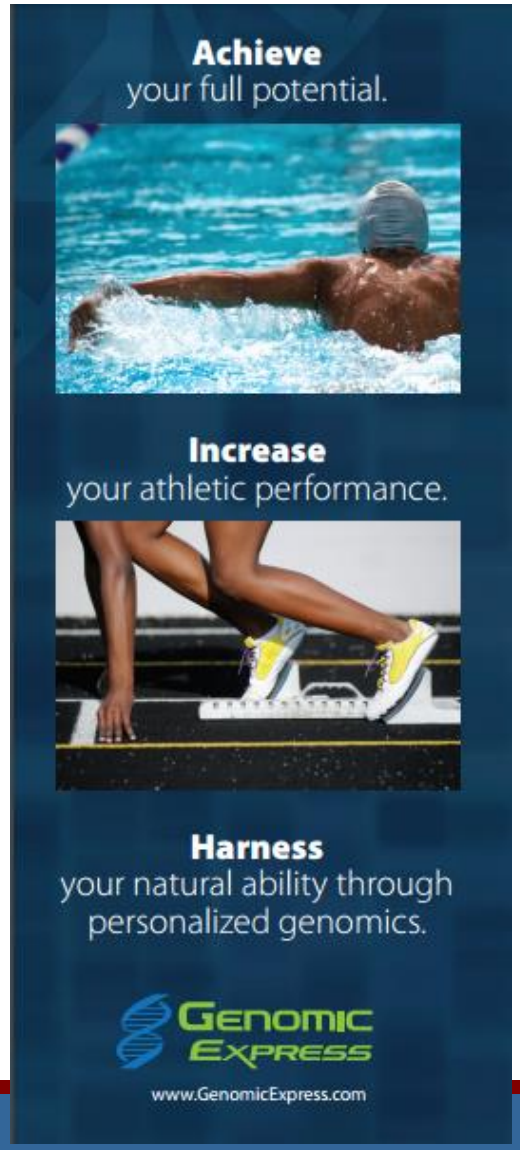
SUPERHERO
DNA Test

NUTRITION
DNA Test

FITNESS
DNA Test

Genomic Testing- Athletic Ability

- Over 36 companies marketing genetic tests
- Poor quality control
- Targeted to coaches and parents
- Individuals also wanting to focus training



Genomic Testing- Consensus Statement

Consensus statement

OPEN ACCESS

Direct-to-consumer genetic testing for predicting sports performance and talent identification: Consensus statement

Nick Webborn,[1] Alun Williams,[2] Mike McNamee,[3] Claude Bouchard,[4] Yannis Pitsiladis,[5] Ildus Ahmetov,[6] Euan Ashley,[7] Nuala Byrne,[8] Silvia Camporesi,[9] Malcolm Collins,[10] Paul Dijkstra,[11] Nir Eynon,[12] Noriyuki Fuku,[13] Fleur C Garton,[14] Nils Hoppe,[15] Søren Holm,[16] Jane Kaye,[17] Vassilis Klissouras,[18] Alejandro Lucia,[19] Kamiel Maase,[20] Colin Moran,[21] Kathryn N North,[14] Fabio Pigozzi,[22] Guan Wang[5]

▸ Additional material is published online only. To view please visit the journal online (http://dx.doi.org/10.1136/bjsports-2015-095343).

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Accepted 25 September 2015

ABSTRACT
The general consensus among sport and exercise genetics researchers is that genetic tests have no role to play in talent identification or the individualised prescription of training to maximise performance. Despite the lack of evidence, recent years have witnessed the rise of an emerging market of direct-to-consumer marketing (DTC) tests that claim to be able to identify children's athletic talents. Targeted consumers include mainly coaches and parents. There is concern among the scientific community that the current level of knowledge is being misrepresented for commercial purposes. There remains a lack of universally accepted guidelines and legislation for DTC testing in relation to all forms of genetic testing and not just for talent identification. There is concern over the lack of clarity of information over which specific genes or variants are being tested and the almost universal lack of appropriate genetic counselling for the interpretation of the genetic data to consumers. Furthermore independent studies have identified issues relating to quality control by DTC laboratories with different results being reported from of the evidence in relation to genetic testing and the limitations of current knowledge. This article reviews the issues around the currently available evidence behind the genetic testing, comments on the ethical considerations and makes recommendations about such tests.

STATEMENT ON BACKGROUND TO THE CONSENSUS PROCESS

A group of world experts in the field of genomics, exercise, sport performance, disease, injury and antidoping gathered with the International Federation of Sports Medicine (FIMS) Scientific Commission for a symposium to discuss the current state of knowledge and to share ideas. One key concern was the misuse of research evidence and the misinformation about genetic testing, particularly when marketed directly to the public, coaches or parents. This is known as DTC testing for the purpose of talent identification and to assess potential for future sports performance. There have been

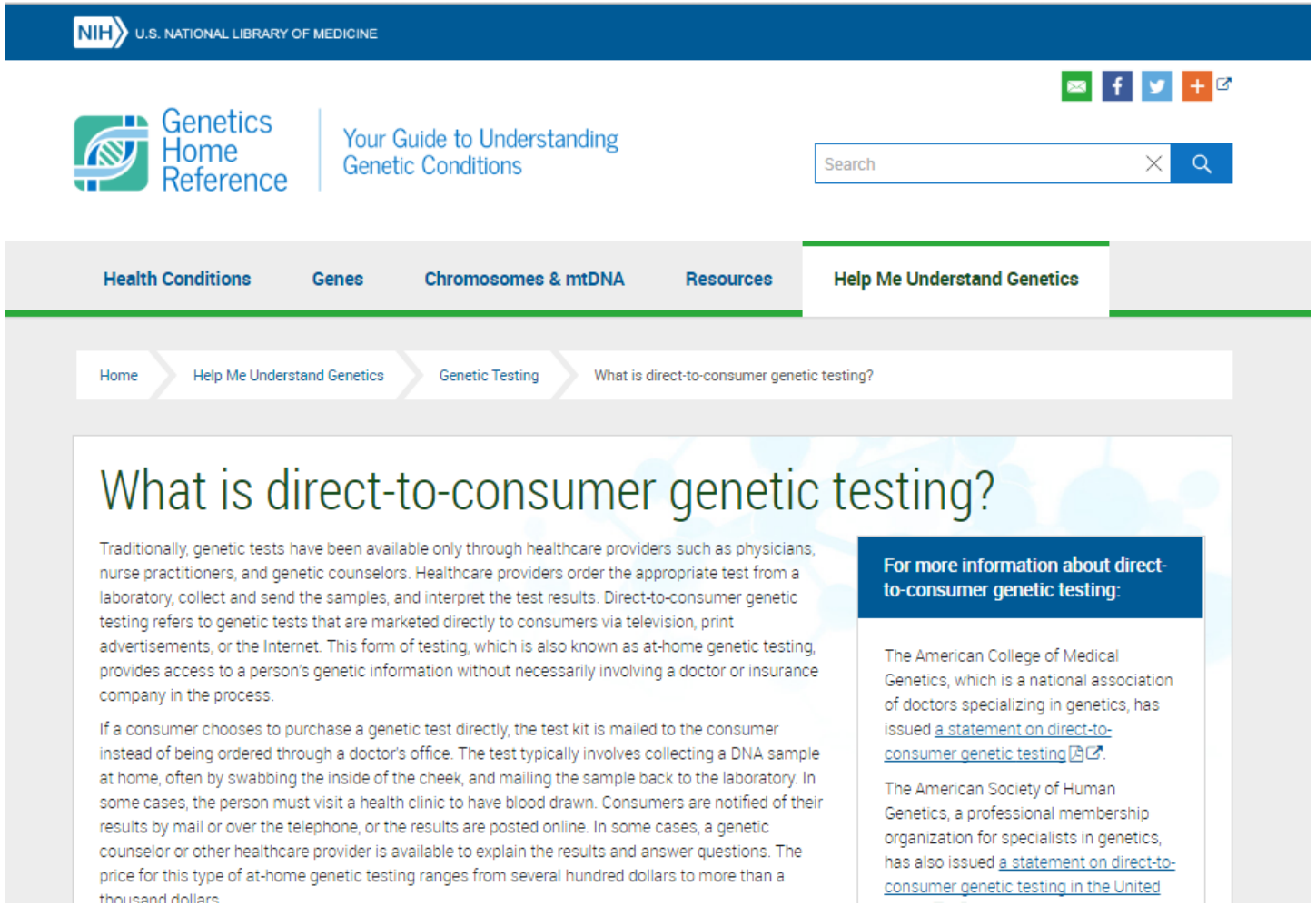
Concerns

- Privacy and legality
- Who has access?
- What all is being done now and in the future with the information?
- Unexpected surprises?
- Test results can vary among companies
- Validity of tests
- No counseling provided

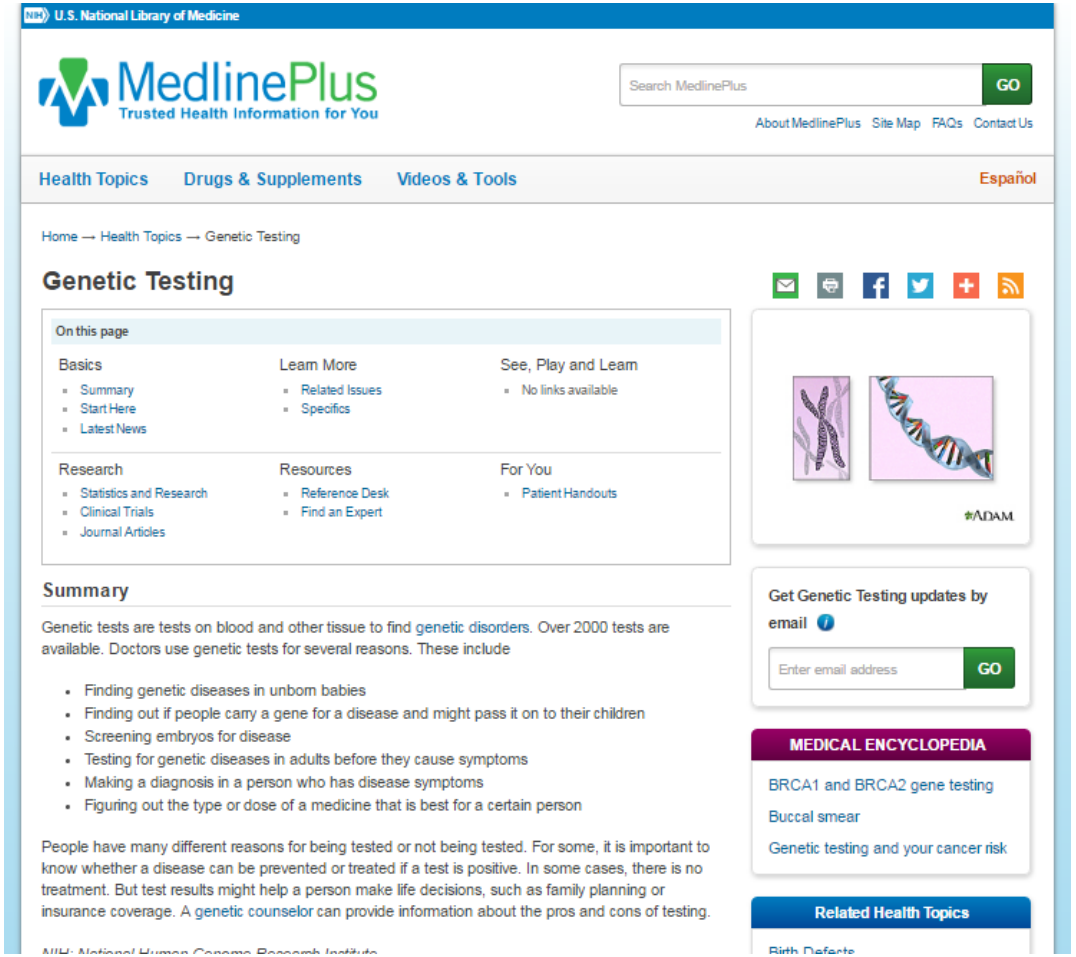
Benefits

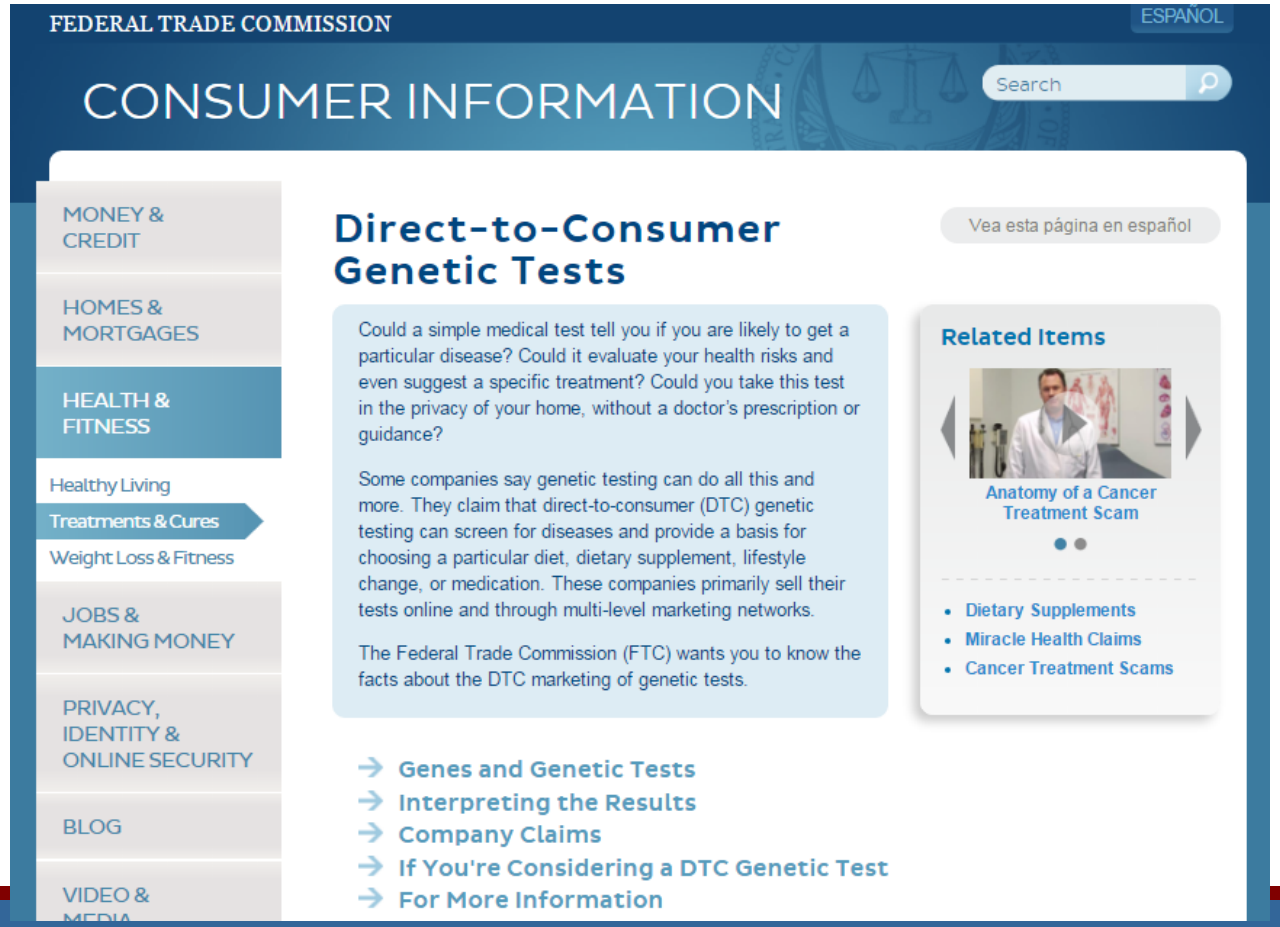
- Learn more about own health
- Learn more about ethnicity and family history
- Bring awareness to family health issues for future generations
- Motivation to work on health habits
- Encourages patient engagement
- Contributing to advancement of healthcare and science
- Moral obligation

Genetics Home Reference

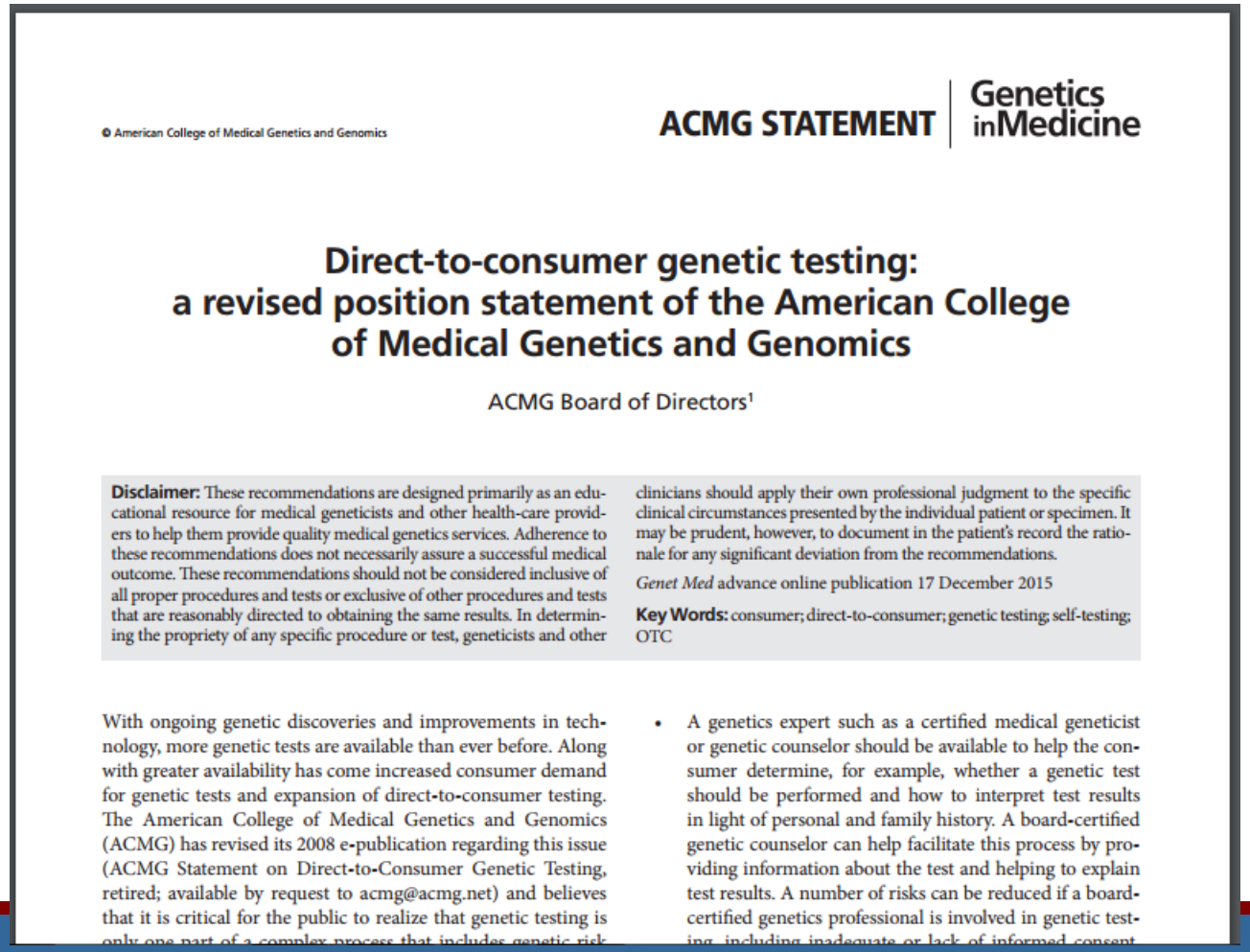


MedlinePlus





American College of Medical Genetics and Genomics



ACMG STATEMENT | Genetics inMedicine

Direct-to-consumer genetic testing: a revised position statement of the American College of Medical Genetics and Genomics

ACMG Board of Directors[1]

Disclaimer: These recommendations are designed primarily as an educational resource for medical geneticists and other health-care providers to help them provide quality medical genetics services. Adherence to these recommendations does not necessarily assure a successful medical outcome. These recommendations should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, geneticists and other clinicians should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from the recommendations.

Genet Med advance online publication 17 December 2015

Key Words: consumer; direct-to-consumer; genetic testing; self-testing; OTC

With ongoing genetic discoveries and improvements in technology, more genetic tests are available than ever before. Along with greater availability has come increased consumer demand for genetic tests and expansion of direct-to-consumer testing. The American College of Medical Genetics and Genomics (ACMG) has revised its 2008 e-publication regarding this issue (ACMG Statement on Direct-to-Consumer Genetic Testing, retired; available by request to acmg@acmg.net) and believes that it is critical for the public to realize that genetic testing is only one part of a complex process that includes genetic risk

- A genetics expert such as a certified medical geneticist or genetic counselor should be available to help the consumer determine, for example, whether a genetic test should be performed and how to interpret test results in light of personal and family history. A board-certified genetic counselor can help facilitate this process by providing information about the test and helping to explain test results. A number of risks can be reduced if a board-certified genetics professional is involved in genetic testing, including inadequate or lack of informed consent

ACMG

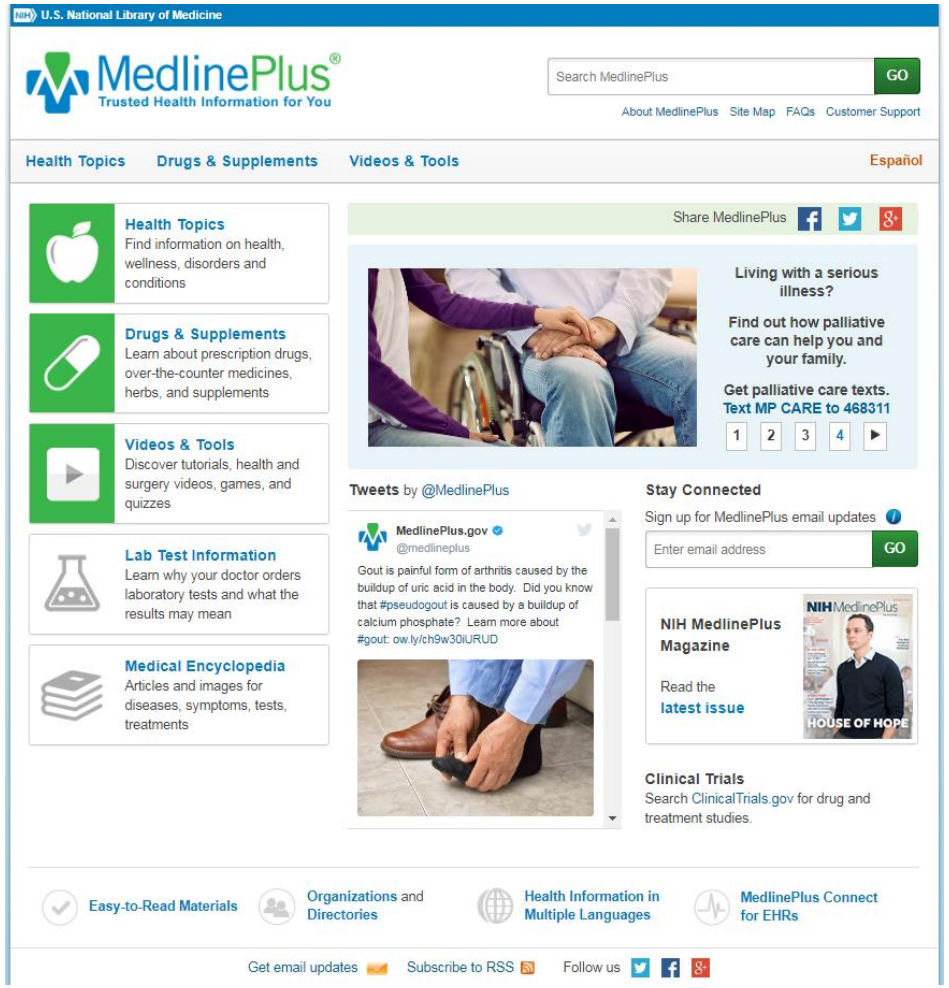
Questions to ask before using a Direct to Consumer Genetic Test

- Is the test right for me?
- What are the company claims?
- What do I hope to find out?
- Am I ready to hear something unexpected?
- Who will the results affect besides me?
- What happens to my genetic information?

Consumer Resources

PATIENT AND K-12 EDUCATION

MedlinePlus



MedlinePlus

- Section: Genetics/Birth Defects
- Health Topic pages:
  - Genetics
  - Genetic testing
  - Genetic counseling
  - Genetic disorders
  - Genetic brain disorders
  - Genes and gene therapy
- text word search

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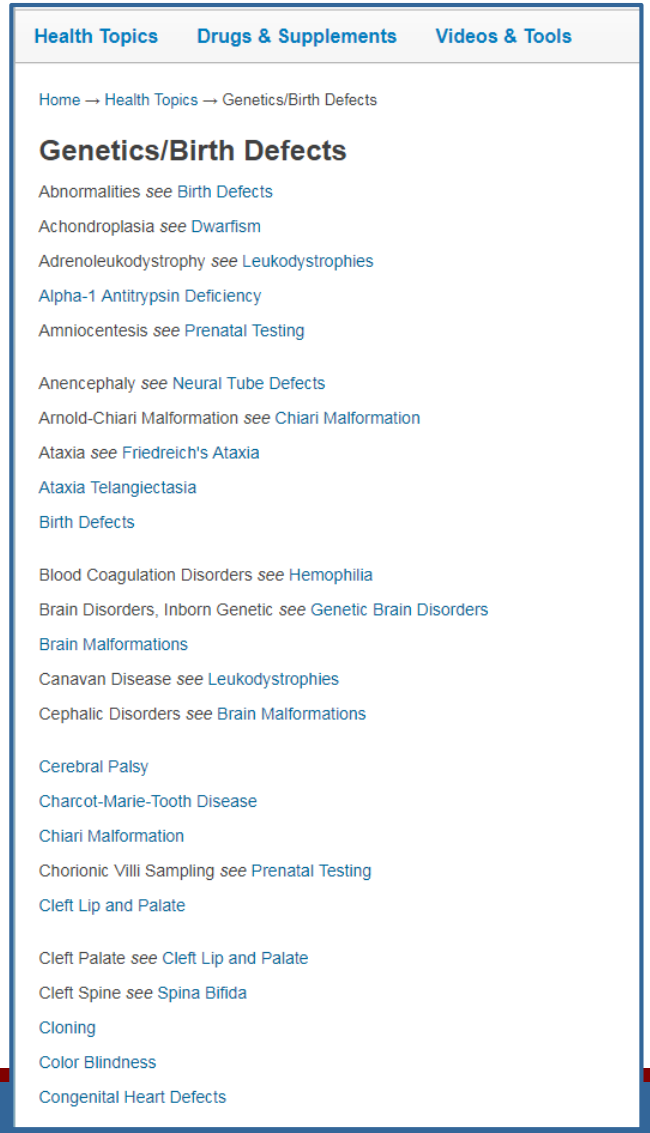
MedlinePlus – Genetics topics



Health Topics | Drugs & Supplements | Videos & Tools

Home → Health Topics → Genetics/Birth Defects

Genetics/Birth Defects

Abnormalities *see* Birth Defects

Achondroplasia *see* Dwarfism

Adrenoleukodystrophy *see* Leukodystrophies

Alpha-1 Antitrypsin Deficiency

Amniocentesis *see* Prenatal Testing

Anencephaly *see* Neural Tube Defects

Arnold-Chiari Malformation *see* Chiari Malformation

Ataxia *see* Friedreich's Ataxia

Ataxia Telangiectasia

Birth Defects

Blood Coagulation Disorders *see* Hemophilia

Brain Disorders, Inborn Genetic *see* Genetic Brain Disorders

Brain Malformations

Canavan Disease *see* Leukodystrophies

Cephalic Disorders *see* Brain Malformations

Cerebral Palsy

Charcot-Marie-Tooth Disease

Chiari Malformation

Chorionic Villi Sampling *see* Prenatal Testing

Cleft Lip and Palate

Cleft Palate *see* Cleft Lip and Palate

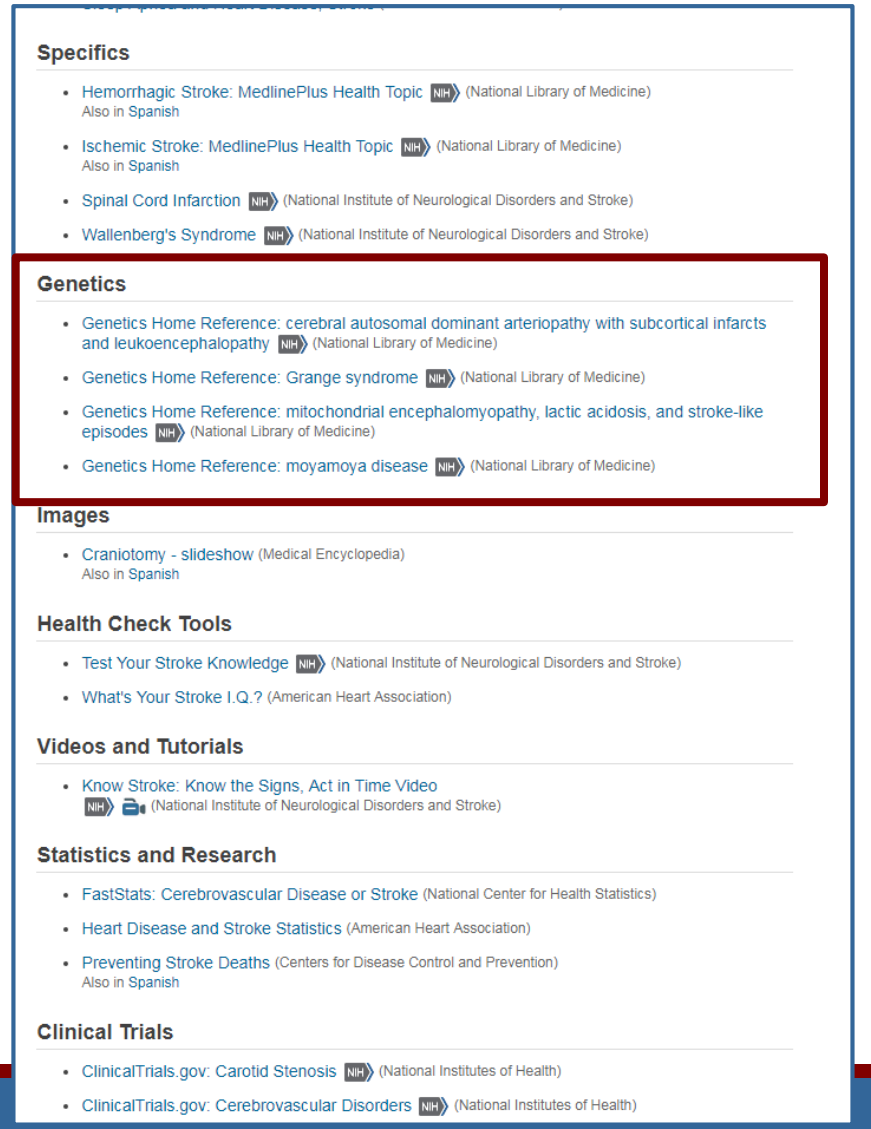
Cleft Spine *see* Spina Bifida

Cloning

Color Blindness

Congenital Heart Defects

MedlinePlus – stroke topic page



MedlinePlus – text search

Textword search 'genetics'

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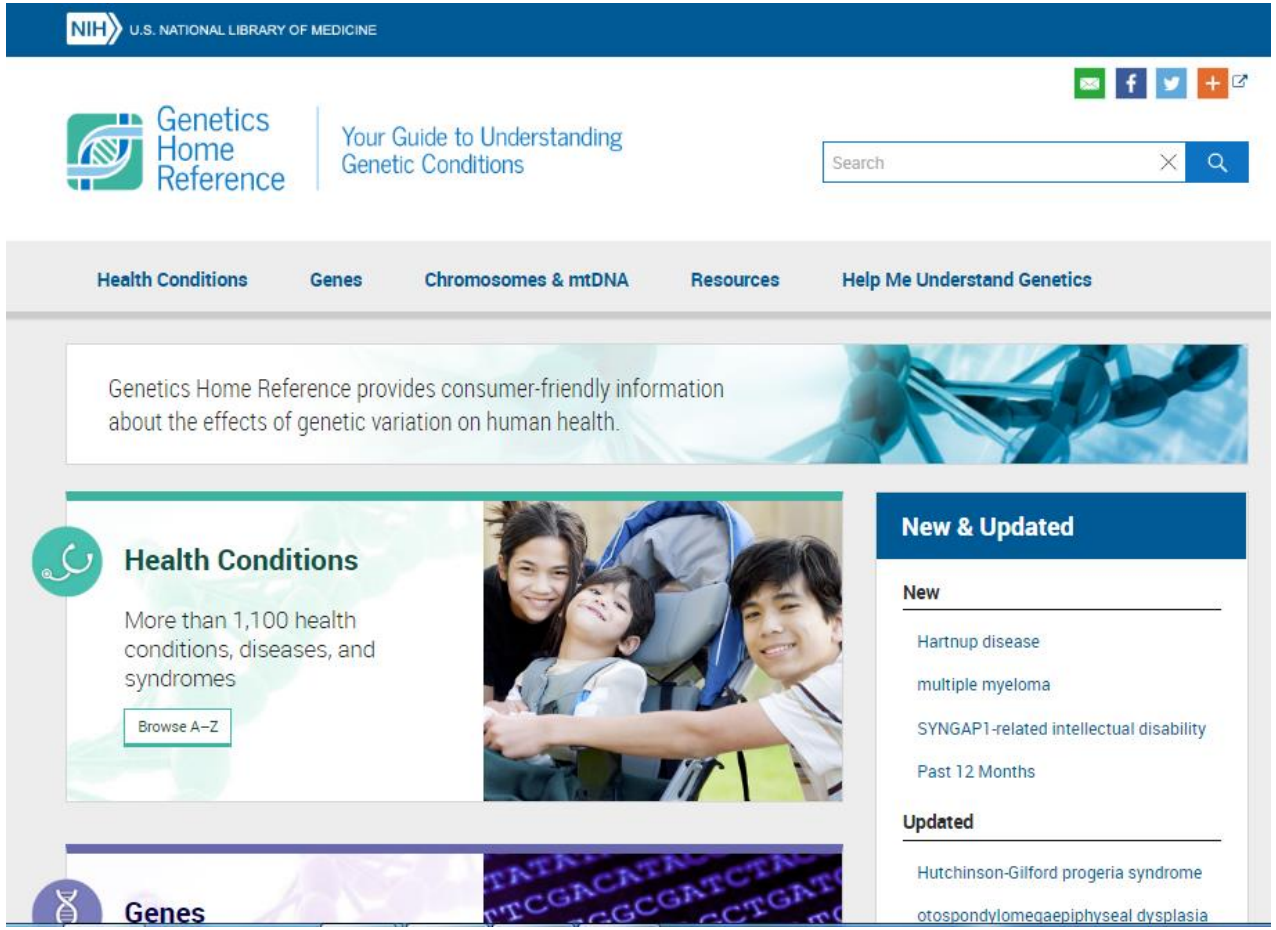
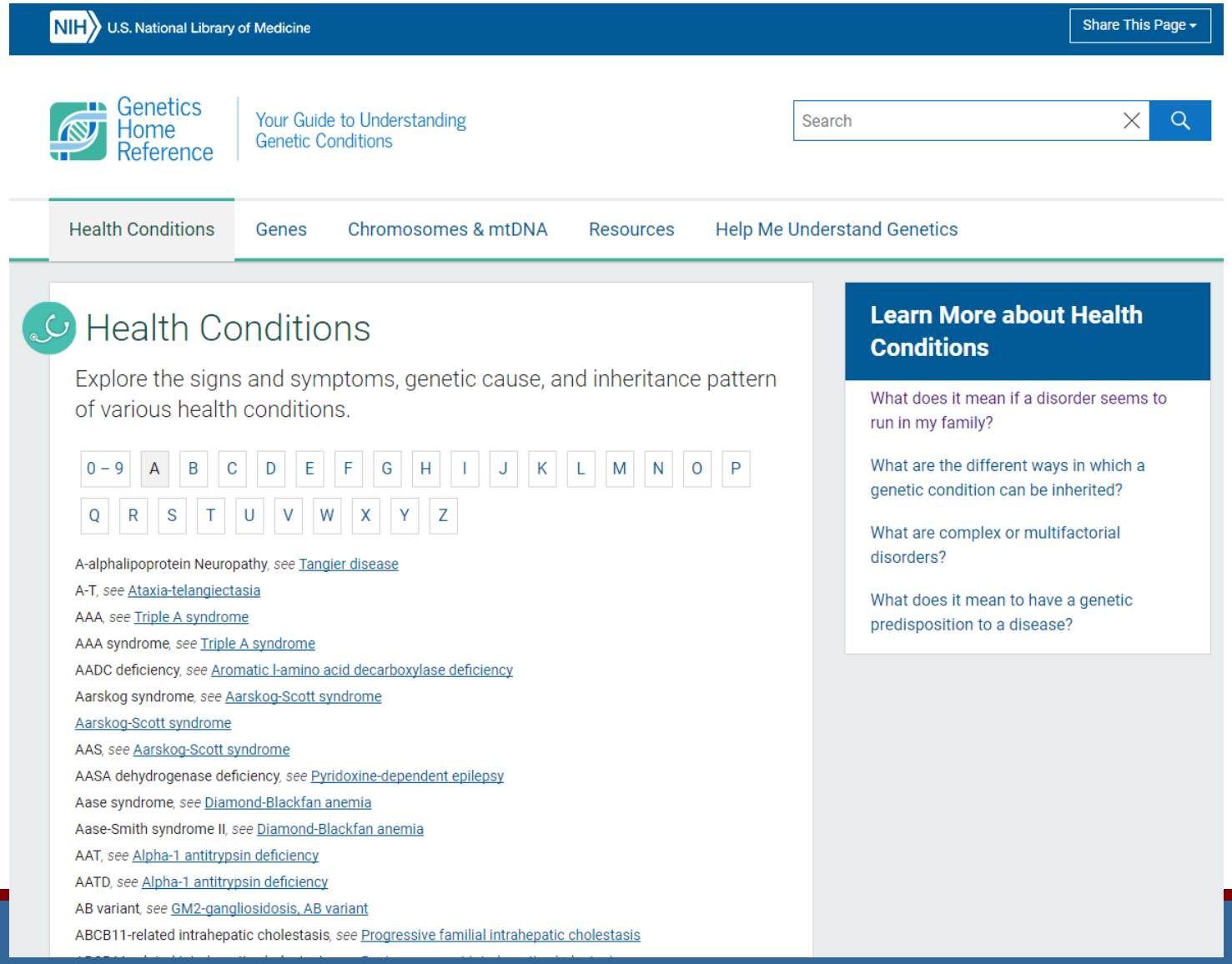


- Health conditions
- Genes
- Chromosomes and DNA
- Resources
- Genetic handbook (Help Me Understand Genetics)

[Genetics Home Reference]

Genetics Home Reference



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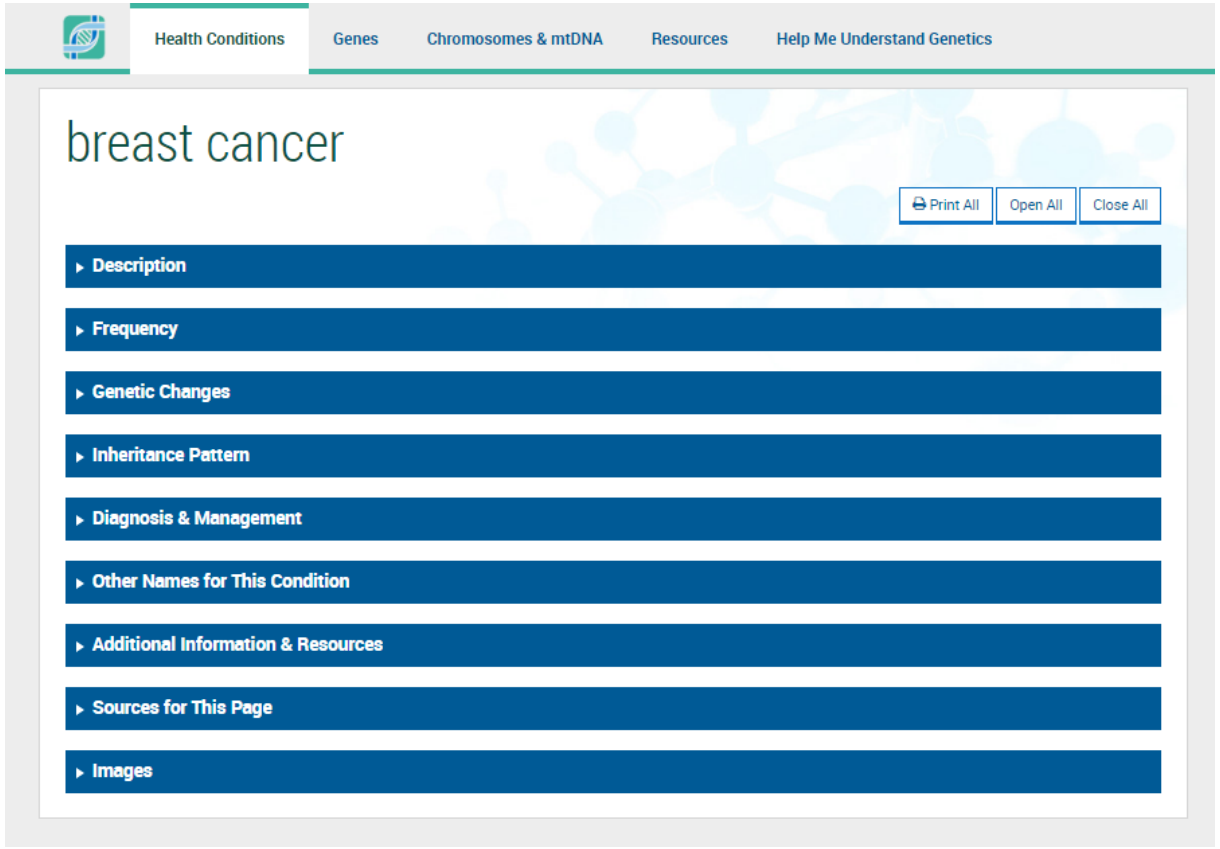
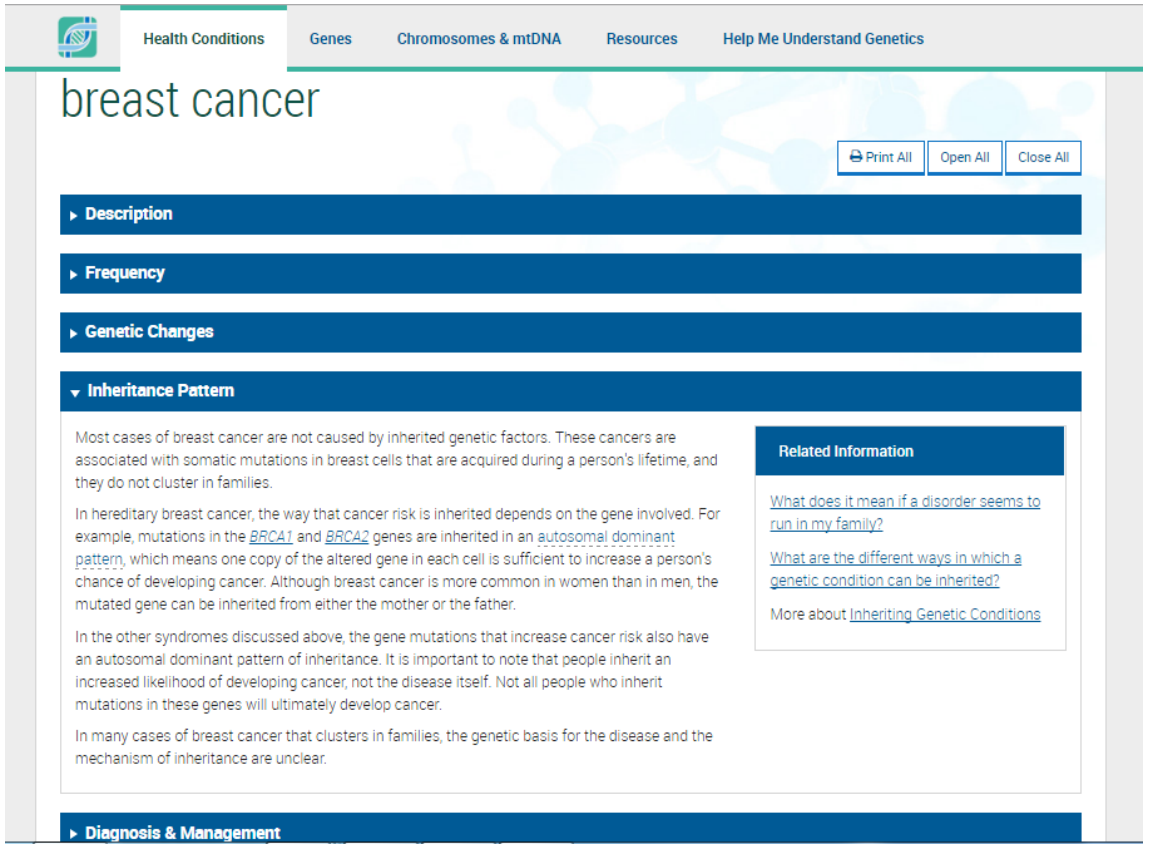
Genetics Home Reference





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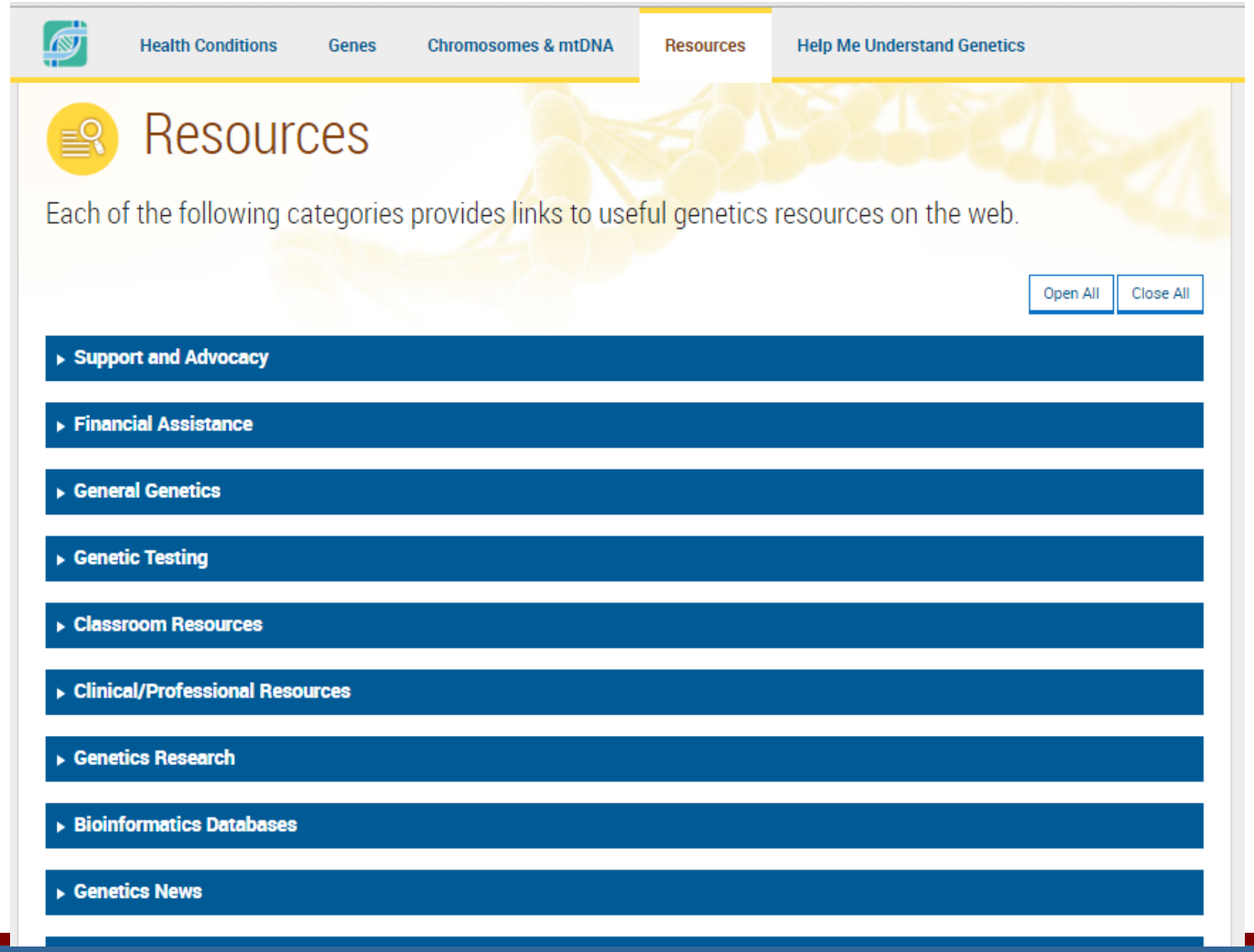

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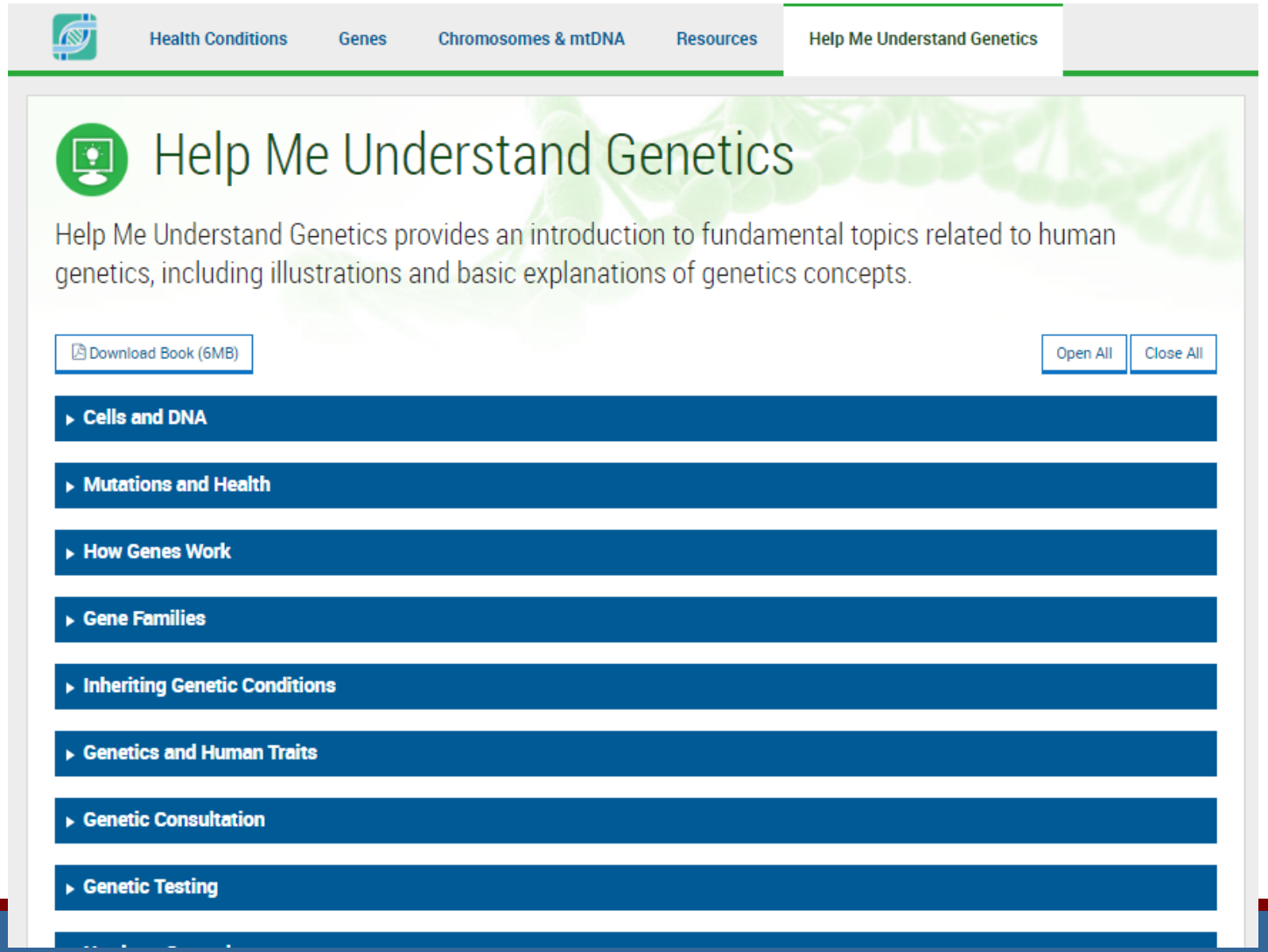

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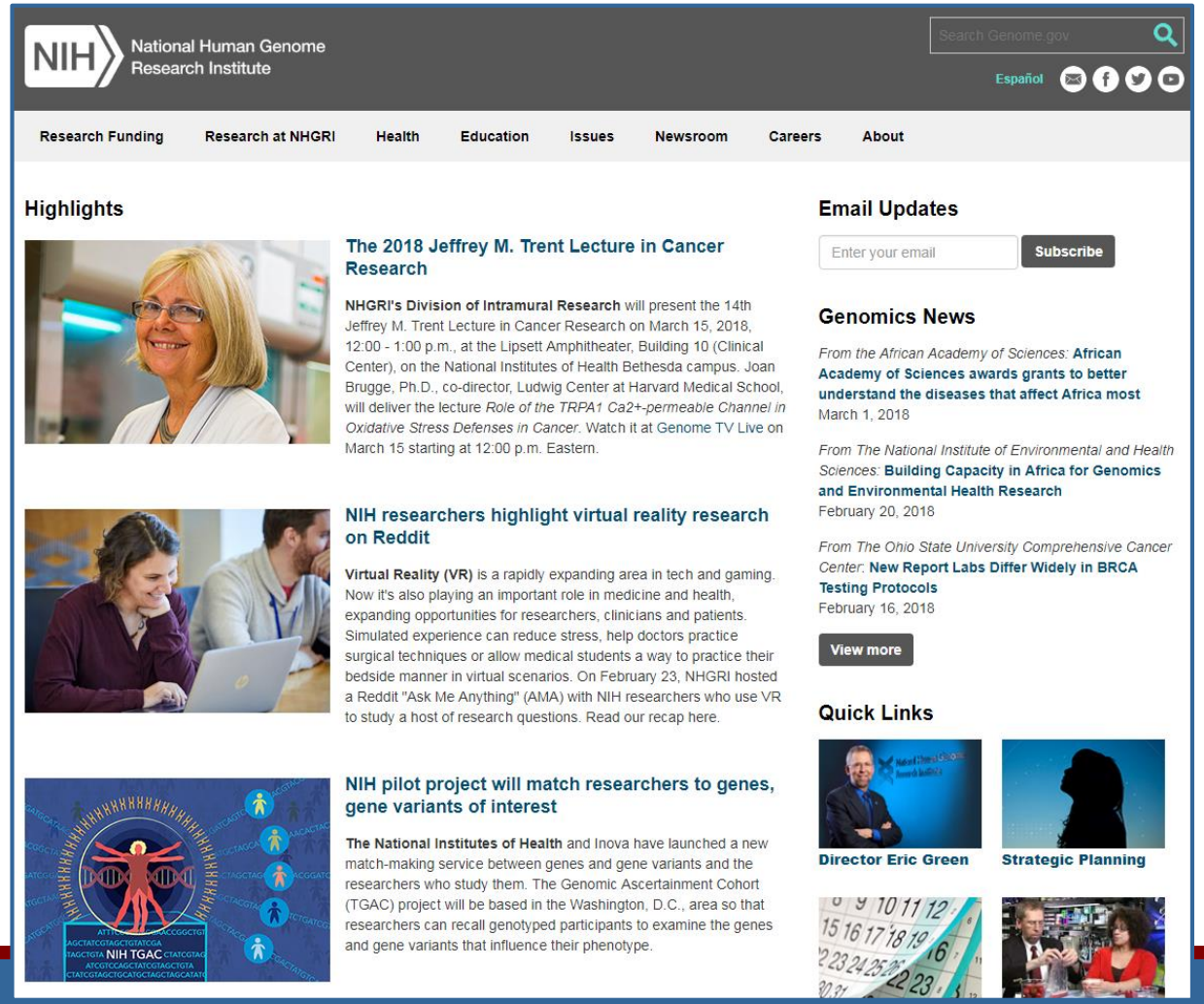
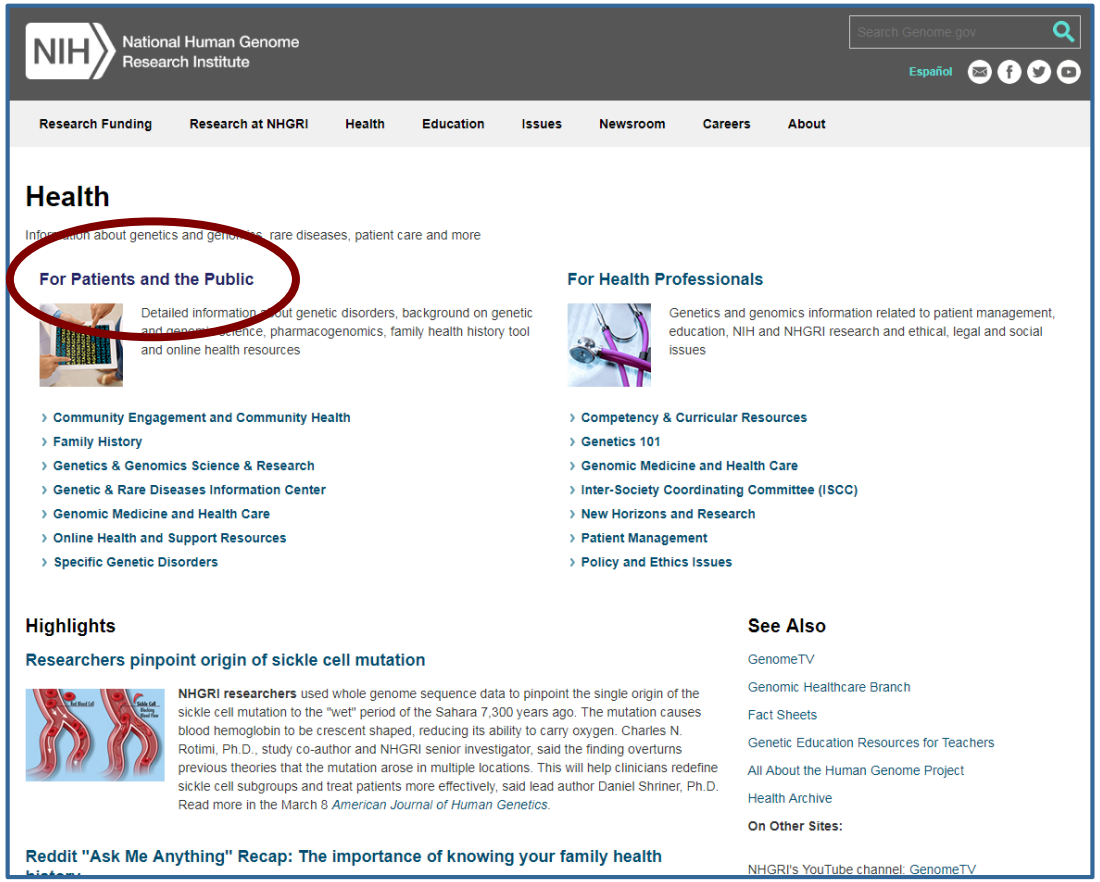
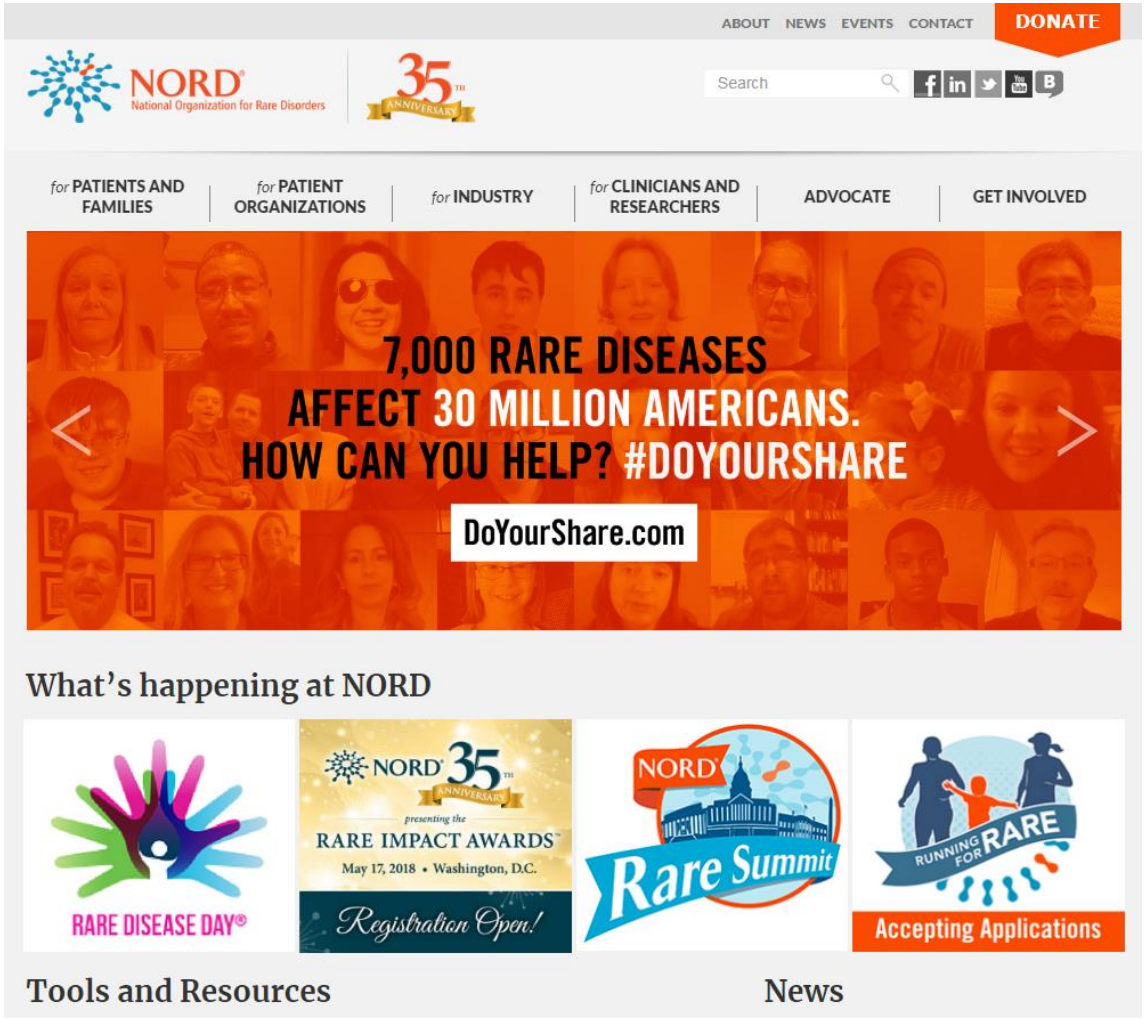
NIH National Human Genome Research Institute- health information



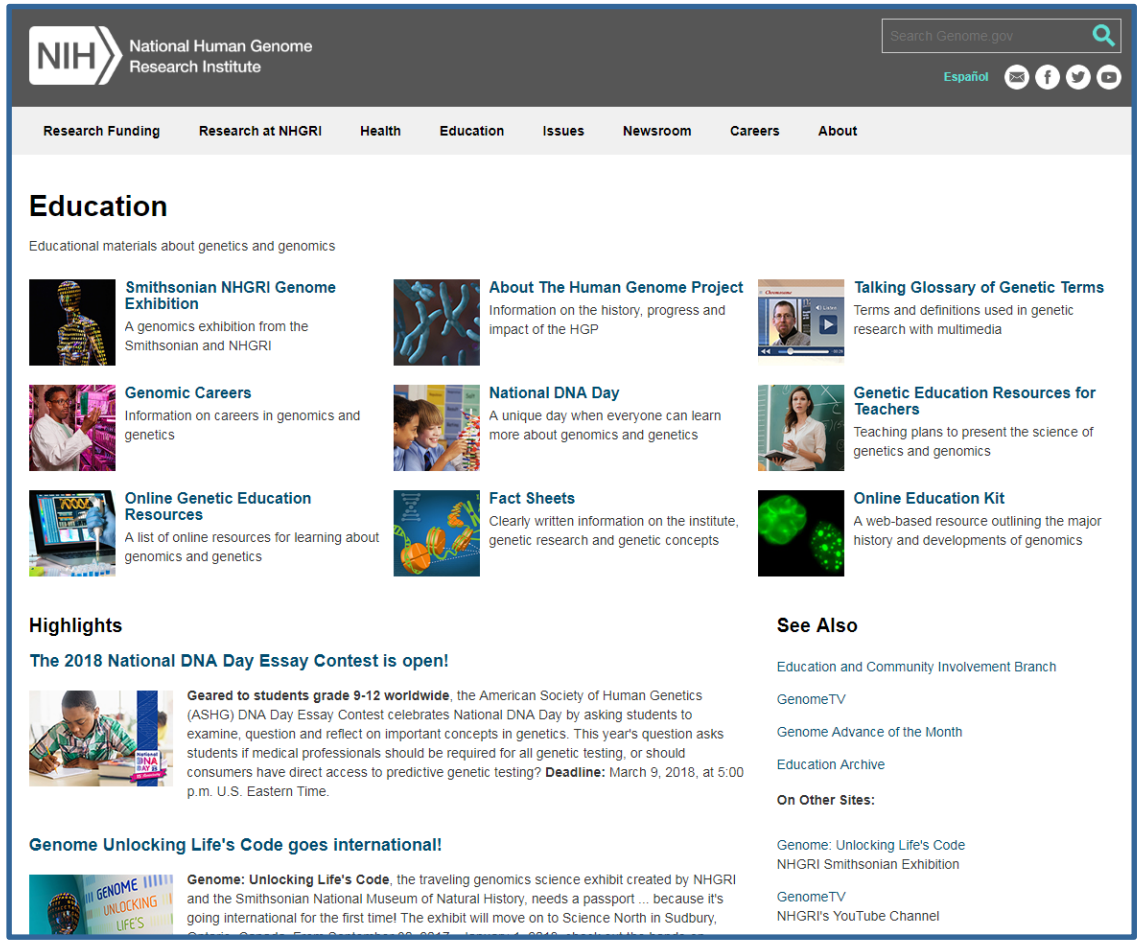


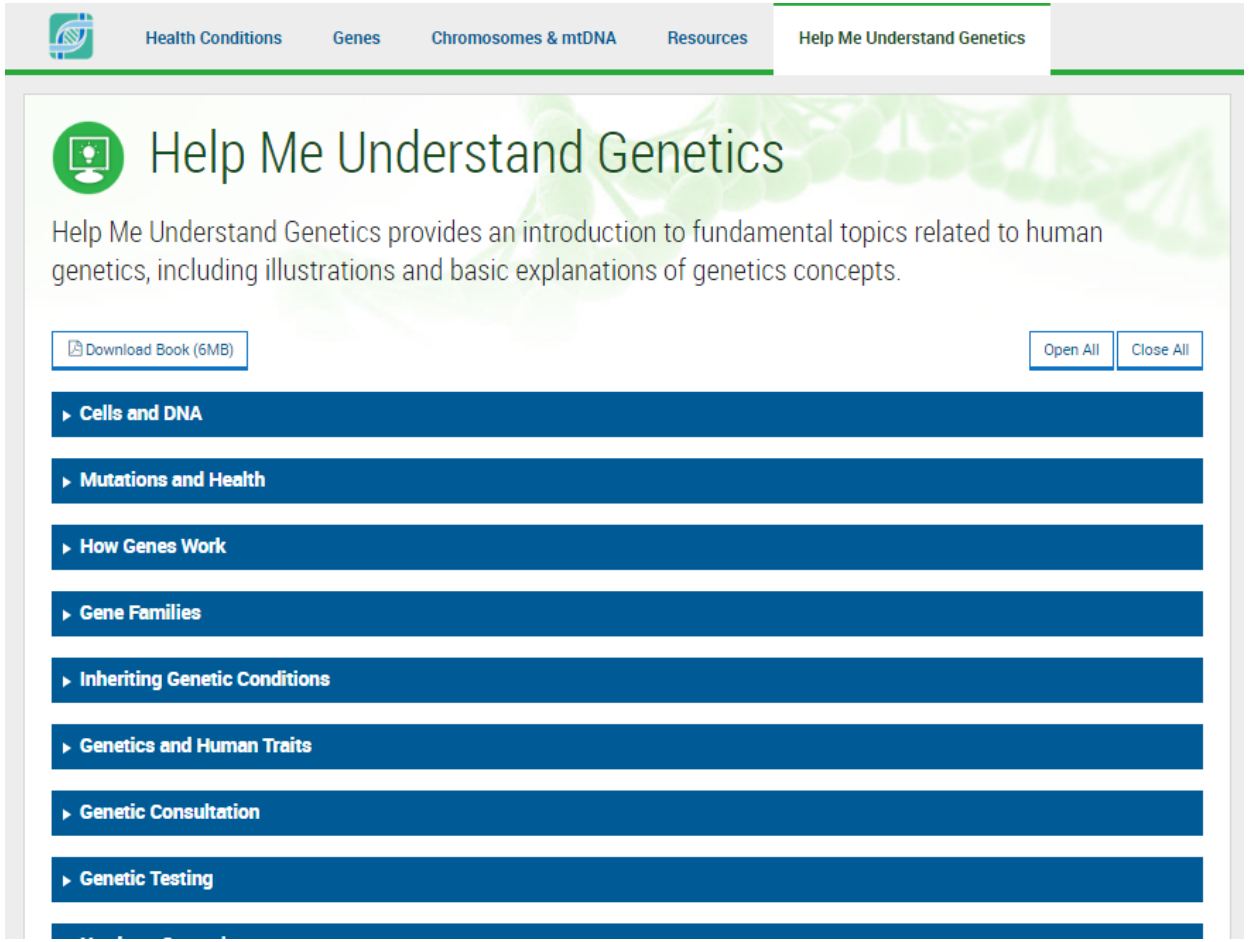
NHGRI

National Organization for Rare Disorders



Education Resources





K-12 Resources

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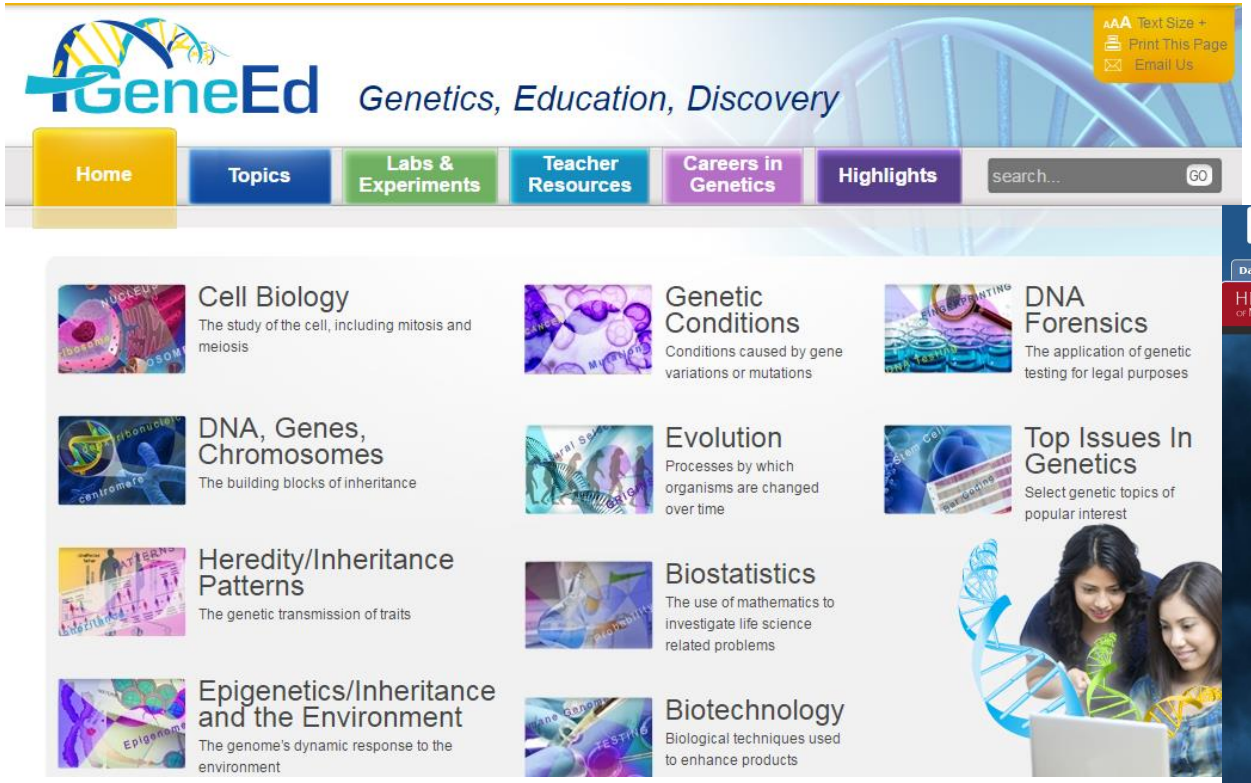
National Network of Libraries of Medicine



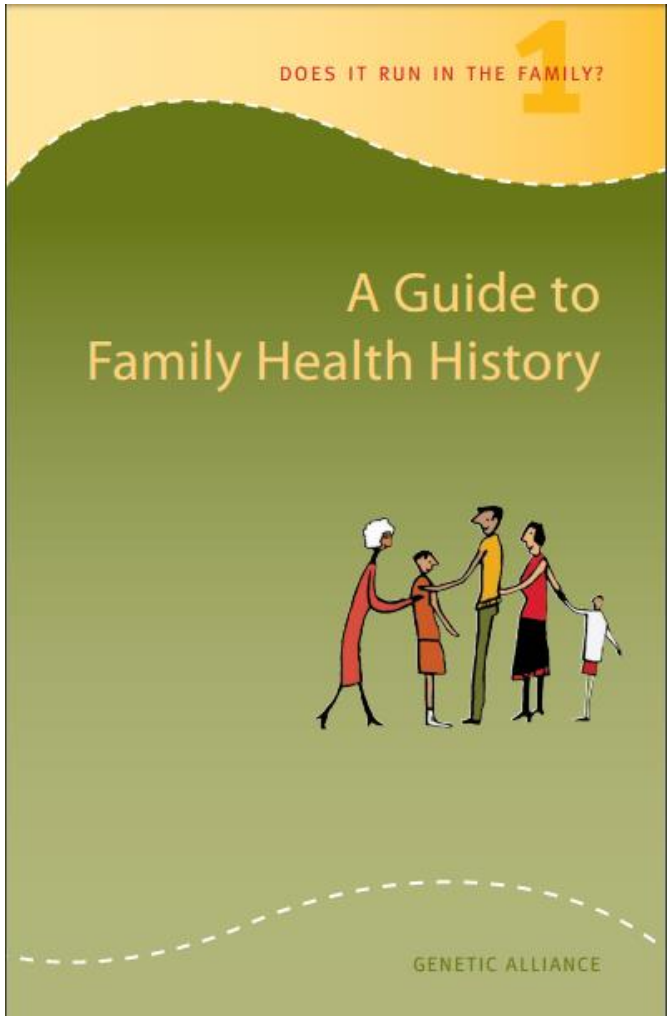
[GeneEd]

[Harry Potter's World]

My Family Health Portrait
U.S. Surgeon General

[Surgeon General's Family Health History Initiative]

Does It Run In the Family? Toolkit



[Does it Run In the Family? toolkit]

Literacy/Education Resources

pgEd Personal Genetics Education Project

ACMG
Translating Genes Into Health®

ASHG THE AMERICAN SOCIETY OF HUMAN GENETICS
discover. educate. advocate.

Genes in Life

GENOME UNLOCKING LIFE'S CODE

GENETIC LITERACY PROJECT
SCIENCE TRUMPS IDEOLOGY

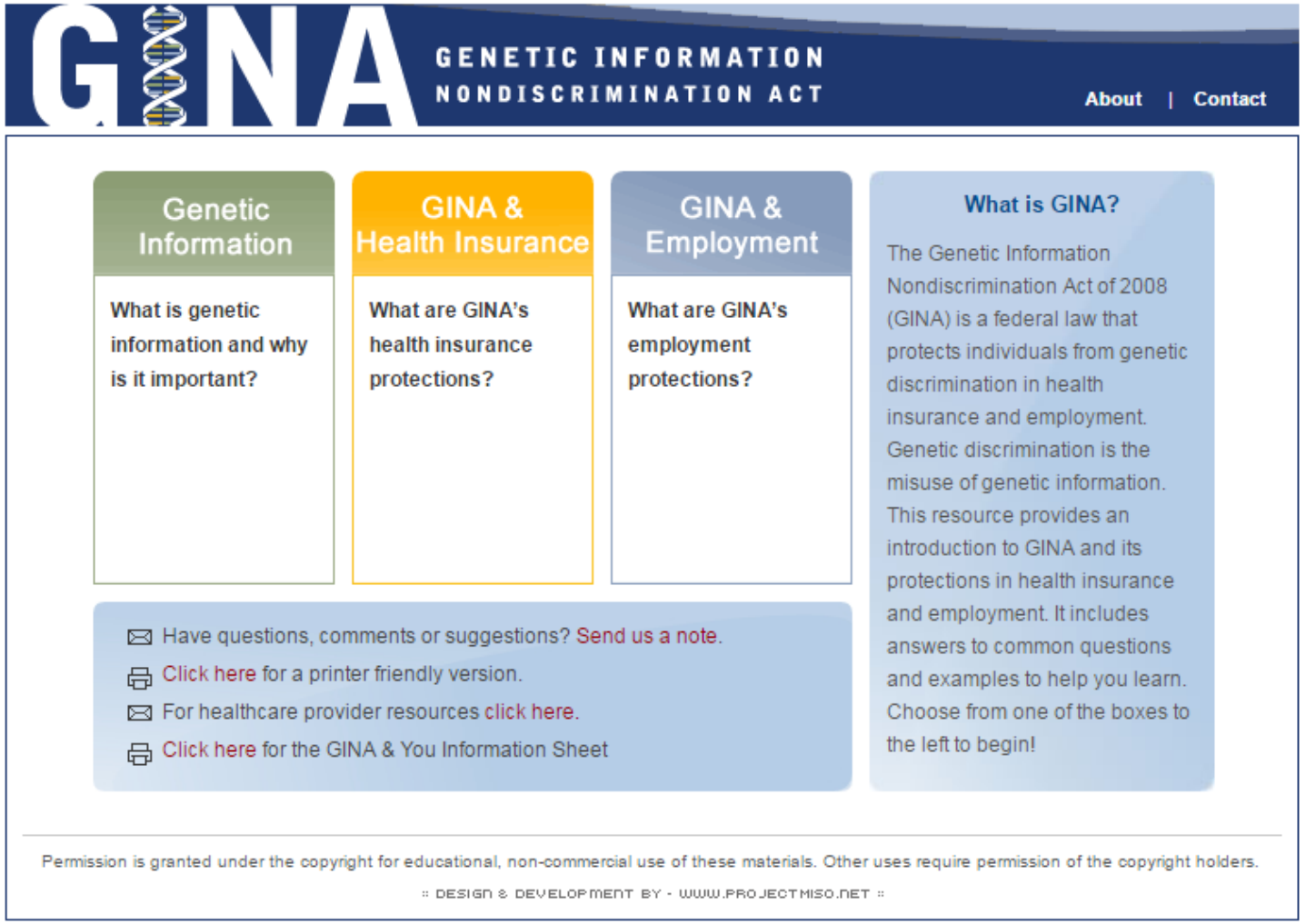
Learn.Genetics
GENETIC SCIENCE LEARNING CENTER

Ethics and Privacy

Societal Concerns

- Who should have access to personal genetic information, and how will it be used?
- Who owns and controls genetic information?
- How does personal genetic information affect an individual and society's perceptions of that individual?
- How will genetic tests be evaluated and regulated for accuracy, reliability and utility?
- Where is the line between medical treatment and enhancement?
- Should testing be performed when no treatment is available?

GINA



GINA Help

H. R. 1313

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H.R.1313 - Preserving Employee Wellness Programs Act

115th Congress (2017-2018) | Get alerts

BILL Hide Overview

Sponsor: Rep. Foxx, Virginia [R-NC-5] (Introduced 03/02/2017)

Committees: House - Education and the Workforce; Energy and Commerce; Ways and Means

Committee Reports: H. Rept. 115-459

Latest Action: House - 12/11/2017 Placed on the Union Calendar, Calendar No. 341. (All Actions)

Tracker: Introduced | Passed House | Passed Senate | To President | Became Law

More on This Bill
Constitutional Authority Statement
CBO Cost Estimates [1]

Subject — Policy Area:
Health
View subjects »

Summary (1) Text (2) Actions (13) Titles (3) Amendments (0) Cosponsors (5) Committees (3) Related Bills (0)

Summary: H.R.1313 — 115th Congress (2017-2018)

All Information (Except Text)

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There is one summary for H.R.1313. Bill summaries are authored by CRS.

Shown Here:
Introduced in House (03/02/2017)

Preserving Employee Wellness Programs Act

This bill exempts workplace wellness programs from: (1) limitations under the Americans with Disabilities Act of 1990 on medical examinations and inquiries of employees, (2) the prohibition on collecting genetic information in connection with issuing health insurance, and (3) limitations under the Genetic Information Nondiscrimination Act of 2008 on collecting the genetic information of employees or family members of employees. This exemption applies to workplace wellness programs that comply with limits on rewards for employees participating in the program.

Workplace wellness programs may provide for more favorable treatment of individuals with adverse health factors, such as a disability.

Collection of information about a disease or disorder of a family member as part of a workplace wellness program is not an unlawful acquisition of genetic information about another family member.

The NEW ENGLAND JOURNAL of MEDICINE

Perspective

JULY 6, 2017

Undermining Genetic Privacy? Employee Wellness Programs and the Law

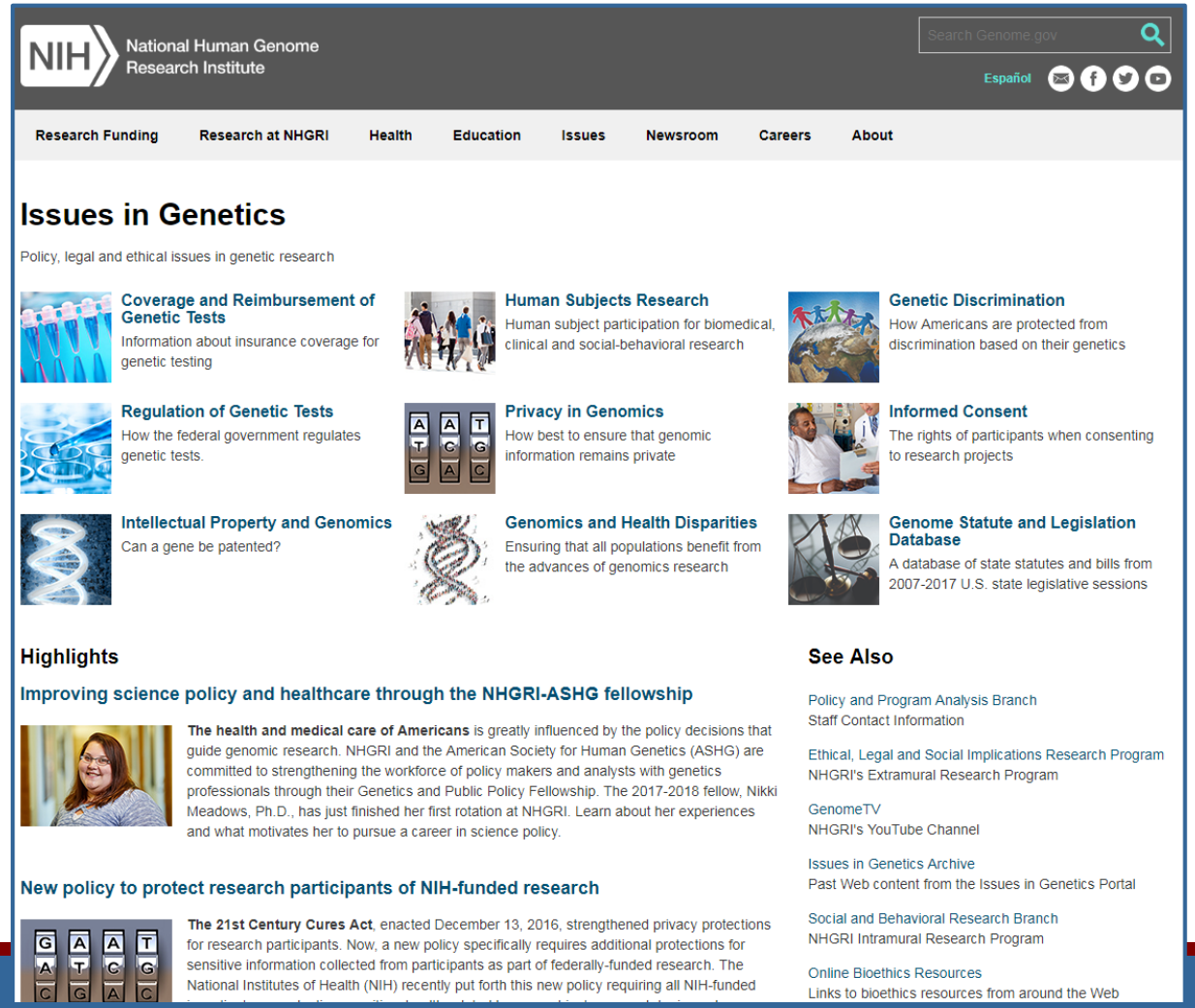
Kathy L. Hudson, Ph.D., and Karen Pollitz, M.P.P.

Genetic information is becoming ubiquitous in research and medicine. The cost of genetic analysis continues to fall, and its medical and personal value continues to grow.

The Genetic Information Nondiscrimination Act of 2008 (GINA) prohibits both employment and health insurance discrimination based on genetic information, and

PMID: 28537794

NIH National Human Genome Research Institute



Informing the Public

CRG Council for Responsible Genetics

Genetic Alliance

All of Us

1 MILLION + VOLUNTEERS

Precision Medicine

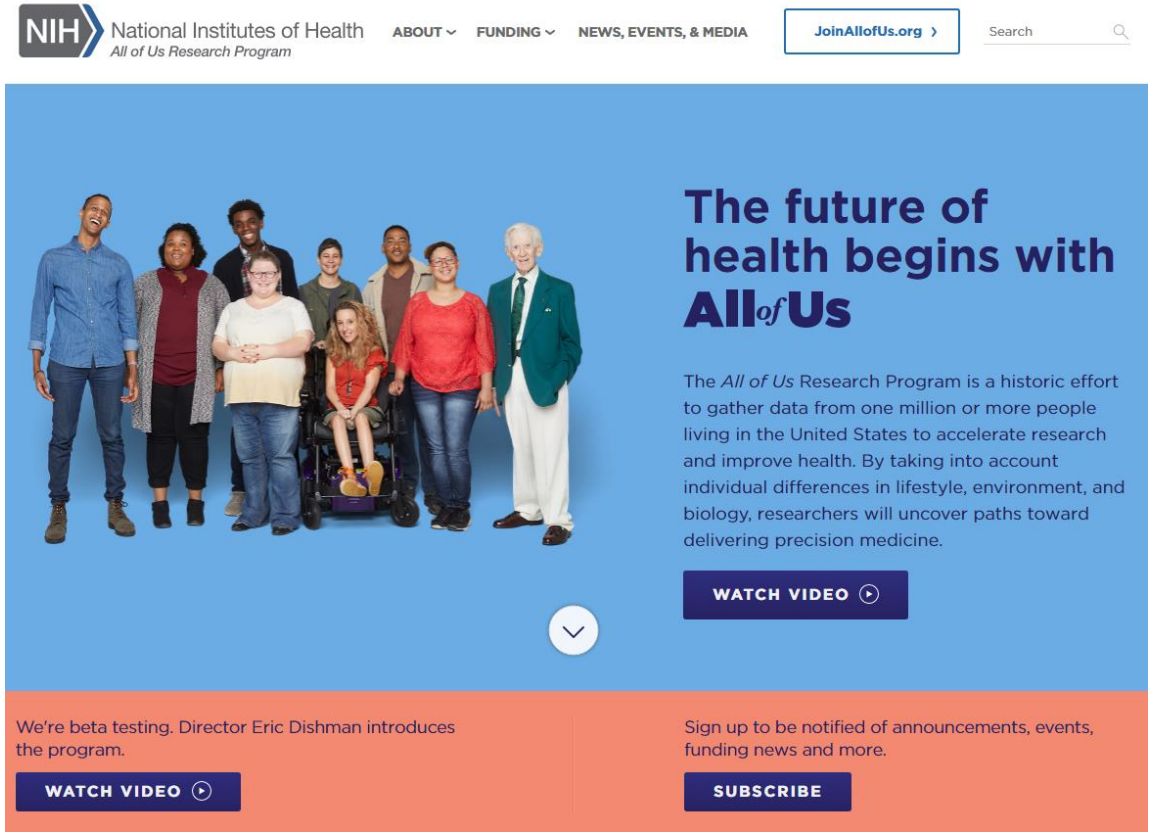
Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.

Instead of what treatment is right for this disease it is what treatment is right for the patient.

Precision Medicine Initiative announcement

All of Us Research Program

The mission of the *All of Us* Research Program is to accelerate health research and medical breakthroughs, enabling individualized prevention, treatment, and care for all of us.

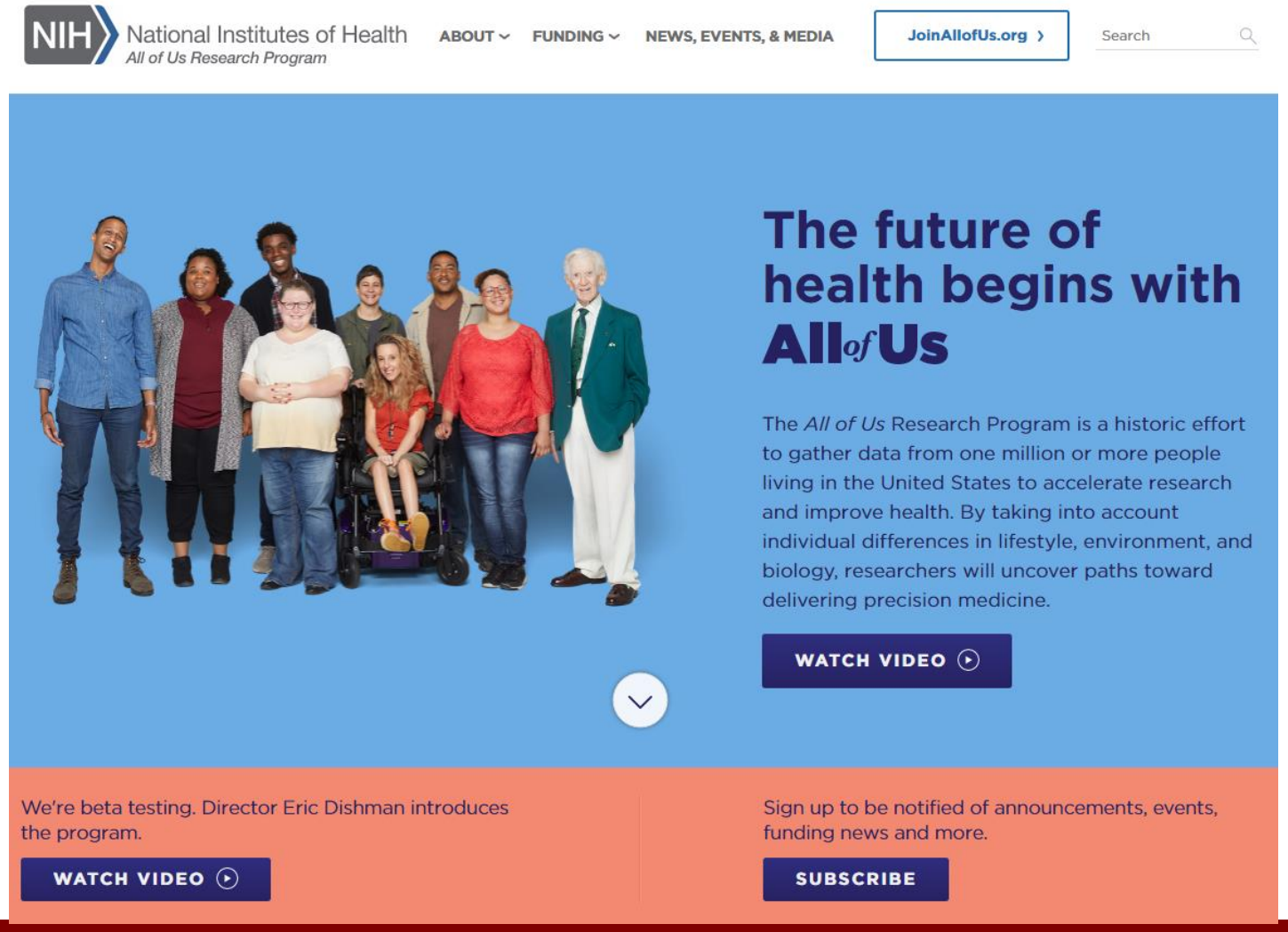


***All of Us* Research Program**

All of Us Research Program- video

What is *All of Us*? video

All of Us – more information



***All of Us* Research Program**

Library role

"Preparing the public to make educated personal and family health decisions in a time of rapidly evolving genetic and genomic knowledge will require new partnerships between the education system, health care systems, the government, community advocacy organizations, consumers and the media."

"What Does it Mean to be Genomically Literate? National Human Genome Research Institute Meeting Report"

Show What You Know!

U.S. National Library of Medicine
National Network of Libraries of Medicine

1. The CDC's top 10 causes of death all have a genetic component.
   True or False?
2. The American College of Medical Genetics and Genomics (ACMG) recommends everyone should use a direct to consumer genetic test.
   True or False?
3. What is the name of the research program that is looking to collect data on 1 million volunteers in order to provide more precise health care through prevention and treatment?
4. GINA (Genetic Information Nondiscrimination Act) protects you from life insurance discrimination.
   True or False?
5. What resource would you recommend to patrons who wanted to learn more about genetic testing?

Thank You!

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